Report of the Australian Genetic Testing Survey 2006

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This document has been prepared for the Royal College of Pathologists of Australasia in consultation with the Human Genetics Society of Australasia. The views expressed in this document do not necessarily represent the views of either organisation.

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Summary

1. **Advances in genetic knowledge have led to the introduction of genetic tests for clinical purposes. Information regarding the current level of testing in Australia would assist in the development of policy and resourcing for such testing.**
2. **In 2007-08, the Royal College of Pathologists of Australasia undertook a survey to document the genetic testing provided in Australia during 2006, with projections for 2007. The survey was funded by the Australian Department of Health & Ageing through the Quality Use of Pathology Program, and involved close collaboration with the Human Genetics Society of Australasia.**
3. **The goal of the survey was to place data about current genetic testing activity in the public domain. This Report summarises the data but does not make recommendations arising from the Survey.**
4. **56 laboratories were identified as providing molecular genetic testing for clinical purposes during 2006 that was not Medicare-rebated (MBS). Data about MBS tests were obtained from Medicare; laboratories were not queried about this activity. 93% of the 56 laboratories provided data for the Survey.**
5. **During 2006, there were five types of MBS molecular genetic tests. A further 437 types of test were offered by Australian laboratories. 55% of these additional types of test were offered by only one laboratory. A further 21% were offered by only two laboratories.**
6. **There were 41,497 assays for MBS molecular genetic tests i.e. 0.07% of all MBS assays. A further 119,354 assays for non-MBS tests were provided. For 75% of the types of test involved, there were less than 100 assays during the year.**
7. **40% of the assays were for medical screening purposes e.g. pre-transfusion testing or neonatal screening. 28% of the assays were for diagnostic purposes. 8% were assays for non-heritable variants in cancer. 5% of assays were to define the genetic status of unaffected relatives in families with a documented mutation. The reason for testing could not be identified in 18% of assays.**
8. **Half of the types of test were provided by laboratories offering less than 10 types of test; 10% of the laboratories offered 40 or more types of test.**
9. **17% of the laboratories reported doing less than 100 assays during 2006; 27% reported doing more than 1,000 assays during this period.**
10. **The majority of types of test were provided by laboratories in only one State or Territory. Only 56 types of test (13%) were provided by laboratories in four or more regions. Some laboratories provided services for patients in other regions, but the rate of testing was higher for samples from within the region than elsewhere.**
11. **28 laboratories (54%) reported that all the types of test they provided were within the scope of their NATA accreditation. Six (11%) reported that none of the types of test they provided were accredited. 18 (35%) reported that they provided a mixture of accredited and non-accredited types of test.**
12. **83% of all types of test were offered only as accredited tests. 4% were offered only as non-accredited types of test. 8% were offered as both accredited and non-accredited types of test by different laboratories.**
13. **The rate of testing for MBS genetic tests varied from 4-fold to over 10-fold across States and Territories. If the non-MBS test data were pooled for each State and Territory, the greatest difference in testing rates between regions was 21-fold. The unequal rates of testing were confirmed on a test-by-test basis.**
14. **The diversity of types of test offered in 2007 increased by approximately 8%. The number of assays rose by 67%, reflecting an increased volume of MBS testing rather than an increase in non-MBS testing.**

1 Introduction

With the continuing "explosion" of genetic knowledge in medicine, there is an increasing gap between the genetic testing that could be provided and the resources that are available. The provision of such resources will probably require support by State and Federal governments, as well as the private sector, and there are a number of models of service provision that could be developed.

There has been a lack of data regarding the current level of demand and supply of genetic testing in Australia. To address this deficiency, the Royal College of Pathologists of Australasia (RCPA), in consultation with the Human Genetics Society of Australia (HGSA), undertook a survey of the genetic testing provided Australia-wide in 2006. The project was funded by the *Quality Use of Pathology Program* of the Australian Department of Health & Ageing, but the Department was not involved in the data collection, analysis, or the production of this report. Dr Graeme Suthers was appointed Survey Coordinator for the project.

Laboratories in the public, academic, and private sectors in Australia were asked to provide details of the type and volume of molecular genetic testing that they provided for medical purposes in Australia during 2006. Medicare-rebated testing was excluded from the request as this information was already in the public domain, and Medicare data are included in the analysis provided below. Testing for research or non-clinical (e.g. paternity testing) purposes was excluded, as was microbial testing for medical purposes. The letter requesting this information, a copy of the questionnaire, and the guideline for completing the Survey are provided in the Appendix to this Report.

The laboratories were not asked to provide any information that might potentially identify a patient or family. Hence there are no privacy concerns in reporting these data. However, the data could be perceived as being commercially sensitive, and the raw data from each laboratory was deemed to be “privileged” and subject to a confidentiality agreement (see Appendix). The tables in the Report provide pooled information, with no details regarding which laboratory in which sector provided the testing.

It is difficult to predict the future demand for testing. While it is obvious that the demand will increase, the demand is closely linked to the level of awareness of testing by both clinicians and patients, the resources available for testing, and the commitment of funders to promote cost-effective testing. Laboratories were asked to provide activity data (actual or predicted) for 2007 to provide a comparison with the "benchmark" year 2006. It was recognized that this represents a short timeline on which to base projections, but the field is changing and growing rapidly, and projections over a longer timeline would not necessarily have been more accurate.

The principle aim of the Survey has been to provide data to inform research and policy discussion rather than to interpret and present recommendations. Hence this Report does not make recommendations about future policy in relation to genetic testing. This Report will be placed in the public domain.

2 The Survey

2.1 Terminology

The Survey sought data both about the types of investigations performed, and the volume of investigations performed. In this Report, the term "tests" refers to types of molecular genetic investigations, and the term "assays" refers to volume of molecular genetic investigations.

The definition of the term "gene" is by no means simple. For the purpose of this survey, a "gene" was defined as a discrete position or locus in the human genome. The great majority of tests

involved interrogation of a genetic sequence which encodes a protein. However, some tests involved interrogation of discrete regions of non-coding DNA.

A test may involve the simultaneous interrogation of multiple genes. Some methodologies currently allow interrogation of 40 or more genes in a single assay e.g. MLPA for micro-deletions which cause intellectual disability. Although highly parallel (or multiplexed) testing represents an efficient approach to molecular genetic testing, the purpose of this survey was to document the diversity of tests provided rather than the efficiencies with which testing was achieved. If the result of a multiplexed assay was a discrete outcome for each gene interrogated, and if the laboratory identified these genes, this was counted as multiple simultaneous tests i.e. one test per gene.

Tests which provided a result for a whole chromosome e.g. assay for trisomy 21, were not included in the Survey. This excluded most cytogenetic investigations with the exception of fluorescent *in situ* hybridisation (FISH) studies directed at a specific gene.

In presenting the results of the Survey, the term "region" refers to an Australian State or Territory.

2.2 Scope of survey

Laboratories were asked to provide information about tests which fulfilled all of the following criteria:

- DNA- or RNA-based testing of human genes for medical purposes.
- Testing for heritable or non-heritable (somatic) genetic variants.
- The samples being tested were collected within Australia.
- The samples were tested during the 2006 calendar year.
- The testing was either performed in an Australian laboratory, or sent from an Australian laboratory to an overseas laboratory (including New Zealand).
- Testing was performed using non-Medicare funds.

The Survey excluded tests which fulfilled any of the following criteria:

- Testing done using Medicare funds.
- Medical testing of non-human genes (e.g. microbial genetic testing).
- Non-medical testing of human genes (e.g. paternity testing).
- Testing done principally for research purposes in relation to a specific project.
- Testing performed on samples received from overseas (including New Zealand).

Laboratories were asked if they wished to offer testing of this gene through an RCPA/HGSA website. This information will be used in the development of an online resource that will assist healthcare professionals and individuals identify laboratories which provide genetic testing, but this information is not provided in this Report.

2.3 Invitations to laboratories, and the responses

There is no single list of laboratories which provide molecular genetic testing in Australia. For the purpose of this survey, a list of laboratories providing molecular genetic testing was developed from the HGSA website[1] (45 laboratories) and the NATA list of laboratories which have been accredited as providers of medical genetic testing[2] (27 laboratories). The directors of research at all Australian universities were asked to provide contact details for any academic laboratories which might have provided testing during 2006, and a further eight laboratories were identified. The Australian Society of Cytogenetics identified seven laboratories. A total of 11 Red Cross offices and laboratories were contacted. A further three laboratories were identified through personal contacts.

In total, 101 letters of invitation to provide data for the Survey were sent. As might be expected, there was some overlap in the mailout, with 22 contacts being identified as redundant. A further 23 laboratories reported that they did not provide testing within the scope of the Survey during 2006.

[1] www.hgsa.com.au
[2] www.nata.asn.au

Responses from the remaining 56 laboratories provide the basis for this Report. It is recognised that there are probably other laboratories providing molecular genetic testing for clinical purposes in Australia. However, the current Survey appears to cover the great majority of the larger laboratories. One challenge for the future is to develop a better method for identifying laboratories which provide such testing.

Some organisations hosted a number of laboratories, raising the question of how a laboratory should be defined for the Survey. Each response detailing tests and assay volume was counted as "one laboratory", and so some organisations were recorded as having a number of laboratories. A total of 39 postcodes were recorded for the 56 respondents to the Survey.

Invitations were initially posted to laboratories in November 2007. Of the 56 laboratories, 52 (93%) provided data. Three laboratories agreed to provide data but did not do so by the deadline (14 April 2008); one laboratory did not respond to repeated invitations.

The distinction between public sector, private sector, and academic laboratories is not always clear. As an approximation, 60% of the 57 laboratories were categorised as being in the public sector, with 20% being in the private sector and 20% being principally academic laboratories.

2.4 Names of test types

Laboratories used a wide variety of names for tests. For reasons of consistency, protein-encoding genes were named using the standard international nomenclature provided by the Human Genome Nomenclature Committee[3]. Fusion genes i.e. abnormal genes resulting from one gene fragment being linked to another, are not listed by the HGNC but were named following the HGNC conventions (as implemented by Gulley et al [2007]). Other tests were provided with non-standard names which are indicated with the suffix "#" in this Report.

2.5 Tests rebated by Medicare

The Survey did not seek any information about the sources of funding utilised in the provision of genetic testing. The majority of types of test presented in this Report were provided with State funds, research funds, or patient charges. However, a few tests were available on the Medicare Benefits Schedule.

Medicare data were sourced from the Medicare website[4]. This source does not indicate the name or location of the laboratory which provided the test. In addition, the data are tabulated in relation to the patient's place of residence, not the laboratory's location. Hence the Medicare data provides no information about laboratory practice. For this reason, these data are excluded from some of the analyses presented below; such instances are indicated.

The Medicare-funded tests that were available by the end of 2007 are listed below. This list constitutes the complete list of genetic tests (Group P7) in the pathology section of the Schedule. The assay volumes are catalogued in this Report by type of test i.e. HGNC gene name, not by item number. For example, item #73309 is an administrative item that involves testing of the same gene and patient group as described in #73308; the assay volumes for these two items were combined in this analysis. In tabulated data presented in this Report, the suffix "[MBS]" has been added to the test name to distinguish tests funded by Medicare from identical tests funded from other sources.

Item #	Description	Test name
73308	Characterisation of the genotype of a patient for Factor V Leiden gene mutation, or detection of other relevant mutations in the investigation of proven venous thrombosis or pulmonary embolism - 1 or more tests. [Previously #65168]	**F5 [MBS]**

[3] www.genenames.org/

[4] Data from http://www.medicareaustralia.gov.au/statistics/dyn_mbs/forms/mbsgtab4.shtml in April 2008. Non-test items such as the patient episode initiation item were not considered in the Report.

73309	A test described in item 73308, if rendered by a receiving APP – 1 or more tests.	**F5 [MBS]**
73311	Characterisation of the genotype of a person who is a first degree relative of a person who has been proven to have 1 or more abnormal genotypes under item 73308- 1 or more tests. [Previously #65174]	**F5 [MBS]**
73317	Detection of the C282Y genetic mutation of the HFE gene and, if performed, detection of other mutations for haemochromatosis where: (a) the patient has an elevated transferrin saturation or elevated serum ferritin on testing of repeated specimens; or (b) the patient has a first degree relative with haemochromatosis; or (c) the patient has a first degree relative with homozygosity for the C282Y genetic mutation, or with compound. heterozygosity for recognised genetic mutations for haemochromatosis. [Previously #66794]	**HFE [MBS]**
73318	A test described in item 73317, if rendered by a receiving APP - 1 or more tests.	**HFE [MBS]**
73320	Detection of HLA-B27 by nucleic acid amplification. Includes a service described in 71147 unless the service in item 73320 is rendered as a pathologist determinable service.	**HLA-B [MBS]**
73321	A test described in item 73321, if rendered by a receiving APP - 1 or more tests.	**HLA-B [MBS]**
73323	Determination of HLAB5701 status by molecular techniques or cytotoxicity assay prior to the initiation of Abacavir therapy including item 71203 if performed	**HLA-B [MBS]**
73300	Detection of genetic mutation of the FMR1 gene by nucleic acid amplification (NAA) where: (a) the patient exhibits one or more of the clinical features of fragile X (A) syndrome, including intellectual disabilities; or (b) the patient has a relative with a fragile X (A) mutation. 1 or more tests	**FMR1 [MBS]**
73305	Detection of genetic mutation of the FMR1 gene by Southern Blot where the results in item 73300 are inconclusive	**FMR1 [MBS]**
73314	Characterisation of gene rearrangement by nucleic acid amplification in the diagnosis and monitoring of patients with laboratory evidence of: (a) acute myeloid leukaemia; or (b) acute promyelocytic leukaemia; or (c) acute lymphoid leukaemia; or (d) chronic myeloid leukaemia; each test to a maximum of 4 tests in a 12 month. [Previously #65280]	**BCR/ABL1 [MBS]**
73315	A test described in item 73314, if rendered by a receiving APP - 1 or more tests.	**BCR/ABL1 [MBS]**
73289	*Chromosome studies, including preparation, count, karyotyping and identification by banding techniques of blood – 1 or more tests*	
73287	*Chromosome studies, including preparation, count, karyotyping and identification by banding techniques of 1 or more of any tissue of fluid except blood – 1 or more tests*	

The last two items in this table refer to cytogenetic tests that lay outside the scope of the Survey; they are included in the list for completeness, and some comparative data of cytogenetic versus molecular genetics assay volumes are presented below.

Items #73308, #73309, and #73311 refer to analysis for a specific variant in the F5 gene or to “other relevant mutations”. In practice, many laboratories also test for specific variants in F2 and MTHFR in addition to the variant in F5. However, this is not necessarily the case and these MBS items were counted only as tests of F5.

2.6 Accredited testing

Laboratories were asked to note whether the test (as performed in the specified patient group [see below]) was included within the laboratory's scope of practice in 2006. NATA evaluates laboratories which provide medical testing against NPAAC standards. If a laboratory meets those standards within a particular field or scope of laboratory practice e.g. genetics, the laboratory is accredited and is given a specified “scope of accreditation”. The laboratory is obliged to validate (on an ongoing basis) each test performed within that scope of accreditation.

A “non-accredited” test could refer to a test provided by an non-accredited laboratory (i.e. no successful NATA assessment), or to a test provided by a laboratory that has a different scope of accreditation (i.e. successful NATA assessment in another field of pathology e.g. haematology), or a non-validated test provided by an accredited laboratory (i.e. successful NATA assessment in genetics, but validation of the specific test is incomplete). The Survey did not differentiate between these three possibilities.

2.7 Patient groups tested

For each test, laboratories indicated the type of patients being tested. The distinction in patient group was restricted to diagnostic testing, family testing, screening, testing for somatic variants, and unknown.

- “Diagnostic” refers to testing of an affected patient (of any age, including prenatal) to determine the genetic basis of the disease.
- “Family” refers to testing of an unaffected person (of any age, including prenatal) who is at increased risk of carrying the mutation on the basis of family history[5]. This will usually refer to testing for a mutation already identified in the family, and includes predictive/presymptomatic testing, and carrier testing.
- “Pharmacogenetic" refers to testing of an affected person for heritable genetic variants to guide choice and dose of drug treatment.
- “Screening” refers to testing an unaffected person who is not recognised as being at increase risk of carrying a mutation. This includes neonatal screening for cystic fibrosis, or screening a patient for pharmacogenetic variants prior to commencing drug therapy.
- “Somatic” refers to testing for non-heritable variants, typically in cancer tissue.

Two additional categories were defined for coding purposes:

- "Supplementary" refers to additional testing (in any patient group) to clarify an initial result
- “Unknown” refers to testing for unknown purposes.

For MBS-rebated tests, the description in the MBS schedule was used to categorise the test. However, the descriptions for HFE and FMR1 testing encompass both diagnostic and family testing, and for these tests the patient group was categorised as "unknown”.

2.8 Methods of testing

Laboratories were asked to indicate the method used for each test. There is enormous variety in both the types of method and the implementation of each method in different laboratories. For this reason, the categorisation of methodologies used in the Survey was simple and intended to be indicative rather than exhaustive:

- “Mutation screen” referred to screening for unspecified variants by a method such as DHPLC, SSCP, DGGE, PTT etc that is recognised as potentially missing sequence variants.
- “Sequencing” refers to sequencing of the coding regions of the gene (and adjacent intronic regions) to identify unspecified variants.
- “Sequencing plus MLPA” referred to sequencing of the gene plus assays for duplication/deletion of exons (or larger re-arrangements) to detect unspecified variants using dosage assays such as MLPA, QPCR, and FISH.
- “Southern” referred to a Southern or Northern blot study.
- “Specific assay/s” referred to any assay for a specific variant . This included testing for one or more specific sequence variants, sizing a specific allele, testing for an abnormality of gene methylation, and screening for deletions. The key feature of this method is that the test focussed on a specific mutation or class of mutations in a gene, and did not search for other mutations in the gene.
- Fluorescent *in situ* hybridisation (“FISH”) is a specific assay but it was listed separately as it

[5] The term “predictive” was used in the Survey, but this carries a specific and more limited meaning in clinical genetic practice and so the term "family" has been used in this Report.

was performed as a cytogenetic investigation (involving chromosome preparations and light microscopy) rather than the usual molecular genetic methods.
- "Segregation study" referred to a study based on the inheritance of genotypes or haplotypes within a pedigree.
- "Sent overseas" referred to samples sent overseas (including New Zealand) by the laboratory.

If the laboratory used multiple methods to test a gene, the test was categorised on the basis of the least sensitive method used.

2.9 Number of assays

Laboratories listed the number of assays performed for each type of test in the different patient groups in their state during 2006 and 2007.

Laboratories also listed the number of assays performed each year in different patient groups where the patient resided in another Australian State or Territory. Testing for overseas patients (including New Zealand) was excluded. The source of interstate samples was not documented.

2.10 Test sensitivity

Laboratories were asked to estimate the sensitivity of the test in the specific patient group i.e. the proportion of all clinically relevant mutations in this gene that would be detected. For recessive disorders, the sensitivity was defined as the proportion of people with the disease in whom both mutations could be identified. For family testing i.e. testing a relative for a mutation already identified in the family, the sensitivity is, by definition, 100%.

2.11 Detection rate

Laboratories were asked to indicate the proportion of patients tested who had an abnormal result. For recessive disorders, this referred to the proportion of people in whom two mutations (homozygous or compound heterozygote) were identified.

3 Release of survey data

The raw data provided by the laboratories was regarded as confidential. These data were not available to the oversight committee for this project, the RCPA, the HGSA, or any State or Federal Government Department. The raw data were handled only by the Survey Coordinator who signed a confidentiality agreement (see Appendix) with most laboratories; some laboratories provided data without requiring a confidentiality agreement. The raw data were destroyed when this Report was completed.

In this Report, the data are summarised and presented on a regional i.e. State and Territory, basis. The data are not presented in such a way that within-region or between-laboratory comparisons can be made. It is recognised that this approach may effectively identify the only laboratory in a particular region which provides a particular service, but this is unavoidable. State funding is a major consideration in the provision of genetic testing, and regional comparisons were an essential component of the Survey.

Some laboratory-based measures are also included in this Report. They represent pooled data in which there is no identifying information (including any indication of region).

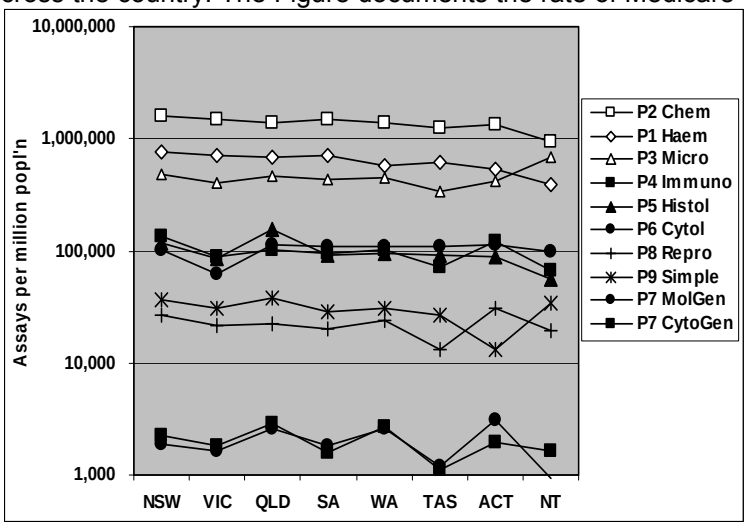
The collated regional and pooled data, including the data underlying figures and graphs in this Report, are tabulated at the end of the Report[6]. These files do not include any identification of the laboratories which provided data.

[6] Electronic copies of this Report (as a WORD file) are available at www.rcpa.edu.au.

4 Medical testing in 2006

During the calendar year 2006, there were 59.5 million pathology assays rebated by Medicare (data in Section 15.1.2). Molecular and cytogenetic assays (Group P7 in the Schedule) accounted for 0.15% of this volume. Cytogenetic assays (items #73287 and #73289) accounted for 52% of the P7 assays i.e. molecular genetic tests that fall within the scope of this Report accounted for just 0.07% of the assays rebated by Medicare during that year.

The rate of testing varied markedly across the country. The Figure documents the rate of Medicare-rebated testing in each of the disciplines of medical testing during 2006. The rates are expressed as tests per million population in each region (population data in Section 15.1.1). Molecular genetic tests and cytogenetic tests have been presented separately. Note that the rate is presented on a logarithmic scale. This highlights that the differences in testing rates across the disciplines varied by three orders of magnitude. Cytogenetic and molecular genetic testing exhibited the lowest rate of utilisation.



However, the logarithmic scale also has the effect of masking differences between regions. Within each discipline, the rate of testing varied by up to 1.7- to 3.3-fold between different regions.

5 Types of test in 2006

Data for this Section are tabulated in Section 15.2.1.

5.1 Types of test

During 2006, 437 different types of test were nominally provided by Australian molecular genetic laboratories. This figure is the number of different types of test for which data were provided for 2006 and 2007.

This figure overestimates the real level of test diversity available during the year. Some types of test were only introduced at the end of the year and should more properly be regarded as being “new tests” that were introduced in 2007. Other types of test were well-established investigations for rare disorders but there were no requests during 2006. In other words, the assay volume for a particular type of test was not necessarily a good guide as to whether the test was an established or new test. Nor did the laboratories always make this distinction clear.

There were 85 types of test (19% of 437) for which there were no assays reported in 2006. Of these, 20 also had no assays reported in 2007. Of the remaining 65 types of test, 61 had 1-10 assays reported in 2007, and the remaining four (<1% of 437) had 11-61 assays. It was likely that these four types of test were new tests in 2007.

In 2007, there were 38 types of test (9% of 437) for which no assays were reported. This included the 20 tests mentioned above for which no assays were reported in either year. Of the remaining 18 types of test, 11 had had small assays volumes (<10 assays per year each) in 2006, and the low level of activity may have simply represented a fluctuation in demand for rare tests. But the remaining seven (1.6% of 437) had had relatively high assay volumes in the preceding year (12-317

assays per year each) and presumably represented the laboratory ceasing to offer the specific type of test.

These differences in assay volume will be considered in more detail below (Section 11), but there has been no attempt to further dissect when tests were introduced during the period 2006-2007 as only modest assay volumes were reported for 1-2% of tests that appeared to have been introduced or withdrawn during the Survey period. Subsequent discussion in this document refers to the 437 tests irrespective of the possibility that some were introduced or ceased during the Survey period.

5.2 Test nomenclature

Almost all (97%) of these tests were investigations of protein-encoding genes or fusion genes. These tests have been catalogued for this Report using the HGNC approved gene name or the derived name (for fusion genes). It is important to note that the HGNC name is not necessarily the most familiar name for a test. However, in the interests of consistency and reproducibility, the HGNC name has been used in this Report. The Table lists some tests as examples of genes for which the HGNC name may not be readily recognized.

Common name	*HGNC name*
AAT	**SERPINA1**
aml-eto	**RUNX1/RUNX1T1**
bcr-abl	**BRC/ABL1**
CHOP	**DDIT3**
CX26	**GJB2**
CX30	**GJB6**
DM	**DMPK**
DRPLA	**ATN1**
E-cadherin	**CDH1**
FRAXA	**FMR1**
GSD1a	**G6PC**
HD	**HTT**
Lamin A/C	**LMNA**
LIS1	**PAFAH1B1**
MCAD	**ACADM**
MEN2	**RET**
MYH	**MUTY**
p53	**TP53**
Rb	**RB1**
SCA1	**ATXN1**
SCA2	**ATXN2**
SCA3	**ATXN3**
SCA6	**CACNA1A**
SCA7	**ATXN7**
tel-aml	**ETV6/RUNX1**
TWIST	**TWIST1**

Tests described as “Prader-Willi/Angelman” (or similar) were coded as being tests of both UBE3A and SNRPN. It is recognized that there are a number of potential targets that could be interrogated on this region of chromosome 15, but specific details were not provided by some laboratories. Similarly, “ANCR” was coded as UBE3A.

Some immunogenetic tests are conventionally described according to the specific DNA variant being sought e.g. HLA-B5701, or HLA-B27. For the purpose of this Report these tests were simply designated as HLA-B tests involving a specific assay rather than being catalogued according to each variant being sought.

Overall, 33% of the tests of protein-coding genes were described with non-standard nomenclature. Attempts were made to ensure that tests with non-standard names were catalogued correctly, but it is possible that errors in assignment were made.

The remaining 13 tests (3%) had non-standard names, and are identified in this Report with “#” as a suffix:

- AZF# assessment of multiple discrete regions on the Y chromosome (no consensus among laboratories re nomenclature);
- Chimerism# assessment of multiple discrete regions (differing between laboratories) to identify mixtures of cells from different people e.g. fetal and maternal cells, or host and donor cells;
- D13S319# assessment of a discrete non-coding DNA region for a deletion indicating cancer prognosis (no HGNC name);
- D19S545# assessment of a discrete non-coding DNA region for a deletion indicating cancer prognosis (no HGNC name);
- D19S851# assessment of a discrete non-coding DNA region for a deletion indicating cancer prognosis (no HGNC name);
- D4Z4# assessment of discrete DNA region associated with a form of muscular dystrophy (no HGNC name);
- D5S721# assessment of a discrete non-coding DNA region for a deletion indicating cancer prognosis (no HGNC name);

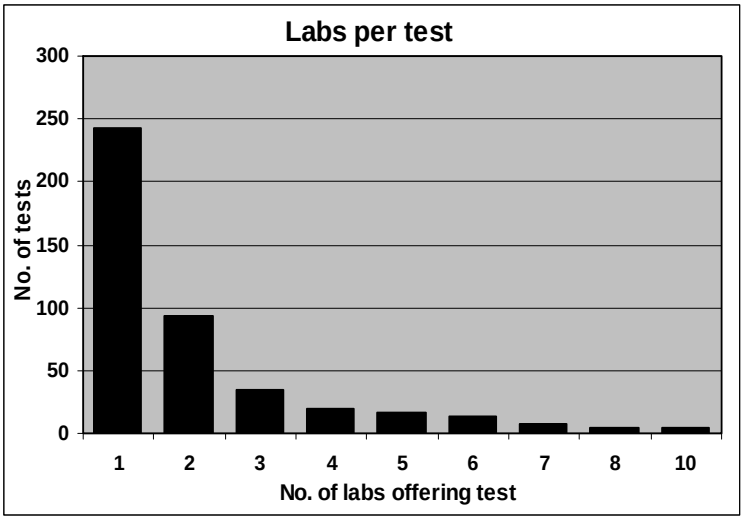
- D7S613# assessment of a discrete non-coding DNA region for a deletion indicating cancer prognosis (no HGNC name);
- MSI# assessment of multiple discrete regions (differing between laboratories) to identify a global characteristic of a type of familial colorectal cancer;
- MT-deletion# assessment for unspecified discrete deletions in mitochondrial DNA;
- Somatic hypermutation# assessment of multiple discrete regions to identify a global characteristic of a type of cancer'
- STR# assessment of multiple discrete regions (differing between laboratories) to identify unspecified deletions or abnormalities of chromosome segregation that carry consequences re a single locus;
- Subtel deletion# assessment of multiple discrete regions (differing between laboratories) to identify unspecified deletions;

Samples that were sent overseas for testing were listed as a single entry rather than being listed for each test requested. The tests done overseas are detailed below (see Section 7.4).

5.3 Laboratories providing type of test

The Figure documents the distribution of the number of Australian laboratories providing a specific test in 2006 (data in Sections 15.1.3 and 15.2.1). Of the 437 tests offered, 243 (55%) were offered by only one laboratory in Australia. A further 94 (21%) tests were offered by only two laboratories nationwide.

Only 5% of genetic tests were provided by more than five laboratories. Note that Medicare-funded testing is excluded from this Figure as Medicare does not provide laboratory-based data.



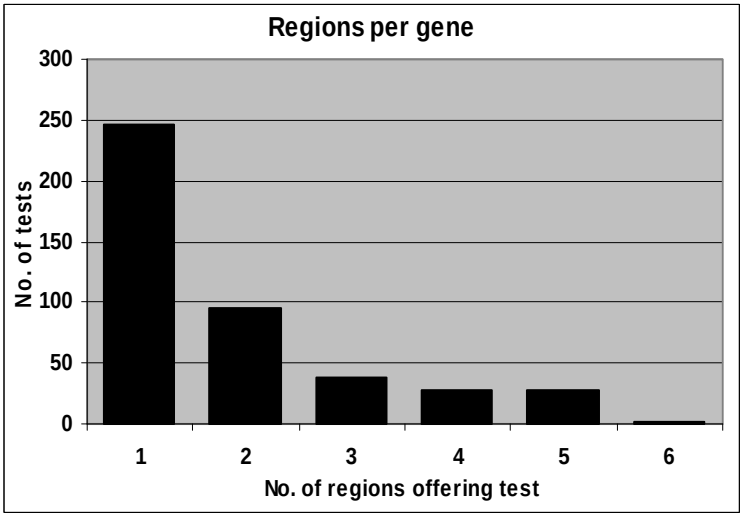
5.4 Types of test per region

The number of types of test offered in each region varied. This does not necessarily imply that access to tests was restricted to that region as many laboratories have the potential to act as *de facto* national laboratories for rare tests. But this issue is considered in more detail in Section 10.

Region	*No. of types of test*
ACT	**4**
QLD	**88**
NSW	**135**
VIC	**171**
SA	**191**
WA	**216**

The Figure summarises the number of regions in which tests were done (data in Sections 15.1.7 and 15.2.1). The majority of tests were provided by one or more laboratories in a single region. Only 56 tests (13% of the total offered) were done in four or more regions. No tests were done in all States and Territories.

Two regions did not report doing any tests (Tasmania and the Northern Territory). Both regions have clinical and laboratory genetic services provided on a contractual basis by other States.



6 Number of assays in 2006

The principle source of data for this section is in Section 15.2.1.

6.1 Assay volume

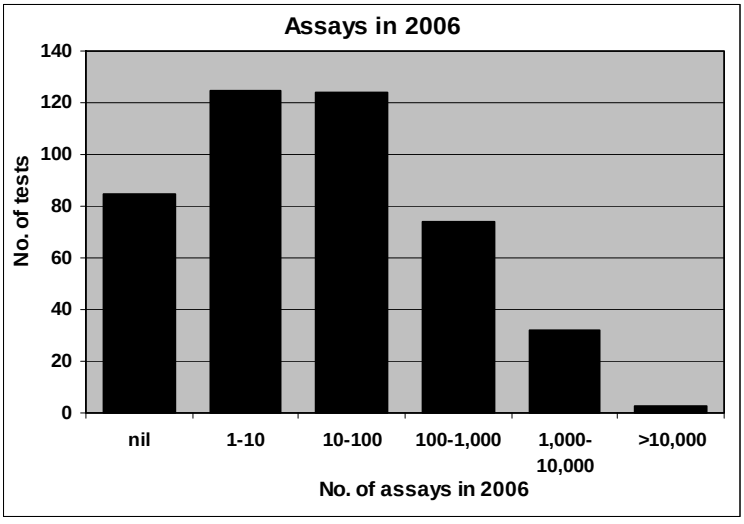
During 2006, a total of 41,497 assays for molecular genetic tests were rebated by Medicare in Australia. As noted above, this accounted for 0.07% of all pathology tests rebated by Medicare in 2006.

In addition to the Medicare tests, a further 119,354 molecular genetic tests were provided by laboratories using non-Medicare funding. The molecular genetic tests provided with non-Medicare funding accounted for 74% of the molecular genetic tests provided overall. The total number of molecular genetic assays (160,851) was equivalent to 0.2% of all pathology investigations rebated by Medicare during the year.

6.2 Assays per type of test

This Figure documents the distribution of the number of assays provided in 2006 per type of test (data in Section 15.1.4). Medicare-funded and non-Medicare-funded testing of the same gene have been counted separately e.g. “F5 [MBS]” and “F5” are counted a separate tests.

There were no assays reported for 85 tests. As noted above (Section 5.1), this could reflect fluctuations in demand for low volume tests, or the introduction of new tests during 2006. The great majority of types of test involved less than 100 assays per year.



6.3 Assays in different patient groups

The Table documents the number of assays performed in different patient groups during the year.

Patient Group	*Assays (n)*	*Assays (%)*
Screening	**64,547**	**40%**
Diagnostic	**45,437**	**28%**
Somatic	**13,092**	**8%**
Family	**7,614**	**5%**
Pharmacogenetic	**470**	**<1%**
Supplementary	**281**	**<1%**
Unknown	**29,411**	**18%**
Total	**160,851**	**100%**

The bulk of the screening assays related to tests for cystic fibrosis (CFTR), immuno-typing (CD109, GP1BA, HLA-A, HLA-B, HLA-B, HLA-C, HLA-DPB1, HLA-DQA1, HLA-DQB1, HLA-DRB1, HLA-DRB3, HLA-DRB4, HLA-DRB5, ITGA2, ITGA2B, ITGB3), and common Jewish mutations (HEXA) (>1,000 assays each).

The most common diagnostic tests were Factor V Leiden (F5), cystic fibrosis (CFTR), thalassaemias (HBA1, HBA2, HBB), haemochromatosis (HFE), α-1-antitrypsin deficiency (SERPIN1A), and sub-telomere deletions causing intellectual disability (subtel deletion#) (>1,000 assays each).

The most common somatic tests were for haematological malignancies: BCL2, BCR/ABL1, IGH@, TRB@, and TRG@ (>1,000 assays each).

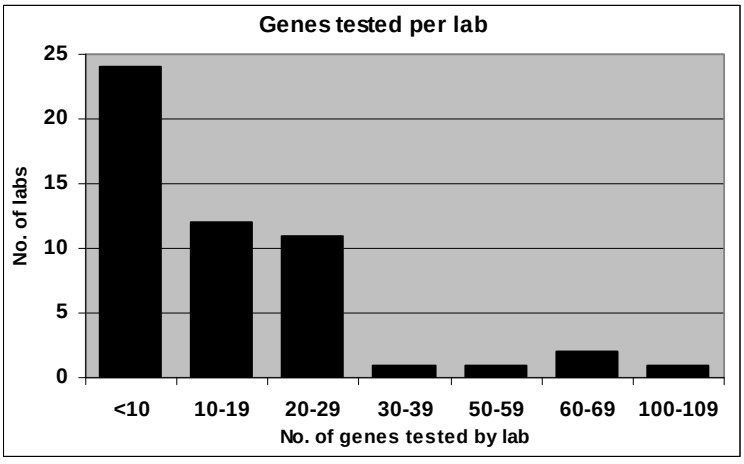
Two of the assays for family members were done more than 1,000 times: cystic fibrosis (CFTR) and Factor V Leiden (F5).

The descriptors for the Medicare items for HFE and FMR1 do not differentiate between diagnostic and family testing. These two tests accounted for the bulk of assays provided to the "unknown" group.

7 Provision of tests

7.1 Types of test per laboratory

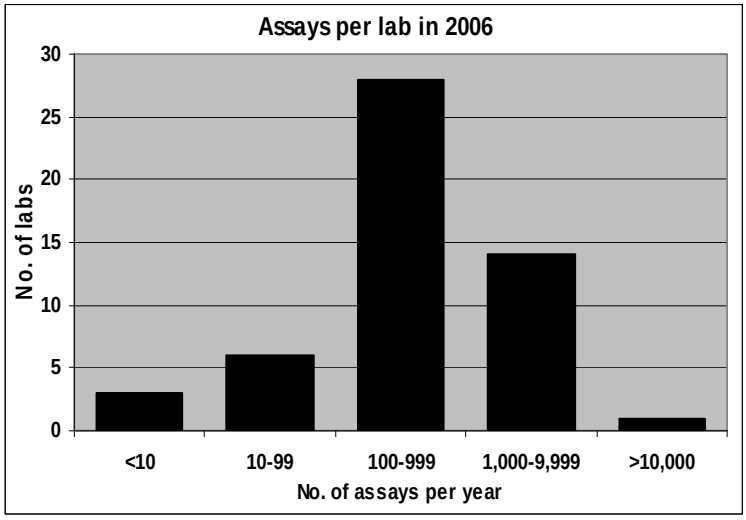
The Figure presents the distribution of the number of laboratories providing a specified number of types of test (data in Section 15.1.5).



The median number of types of test offered by a laboratory was 10, but the range was one to 103. Almost half of the laboratories offered less than 10 types of test. Only 10% of laboratories offered 40 or more types of test. Note that a panel of investigations on a single sample was counted as one assay for each gene included in the panel (see Section 2.1).

7.2 Assays per laboratory

The Figure documents the distribution of the number of laboratories providing a specified number of assays for all tests offered during the year (data in Section 15.1.6). Medicare-rebated tests are not included because those data were not laboratory-based.

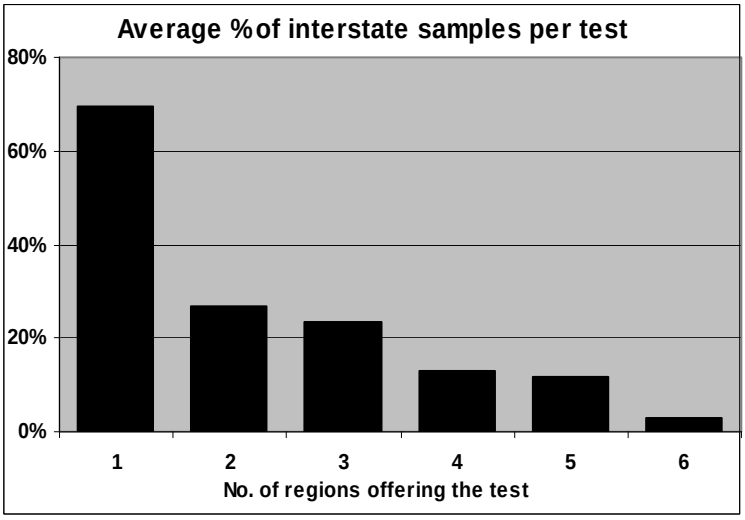


The median number of assays done by a laboratory was 424 per year, but the range was two to 44,150. Three laboratories reported doing a total of less than 10 assays during 2006; in each case, there was at least one other laboratory providing the same type of test in Australia in 2006. Most laboratories did more than 100 assays per year.

7.3 Assays for interstate patients

Testing involved samples from other regions ("interstate samples") for 195 tests (45% of the total of 437 tests). A total of 6,941 interstate samples were tested: this is equivalent to 9% of the intra-region assays for those tests, and an average of 35.6 samples per test for which interstate testing was done (data in Section 15.2.1).

As might be expected, the interstate assay volume was greater for the types of test that were offered by a limited number of regions. If a test is already available in the patient's region, it is less likely that the sample would be sent interstate for testing. The Figure documents the distribution of the average proportion per type of test of assays done on interstate samples versus the number of regions in which the type of test was offered (data in Section 15.1.8).



However, this analysis is limited to those tests for which assays on interstate samples were reported. There were a further 242 tests for which no assays on interstate samples were reported. If the interstate samples reported are apportioned across all tests offered, the proportion of interstate samples per test is much lower. For example, 247 tests were available in only one region (see Section 5.4). A total of 13,881 assays for these tests were performed on intrastate samples during 2006. Only 747 additional assays (an additional 5%) were reported for these tests on interstate samples. This is far lower than might be expected for tests that are only available in one region of Australia. For example, the most populous State (NSW) had 33% of Australia's population in 2006. If there was equivalent access to testing for rare disorders in all regions of Australia, then at least 67% of the assays provided by a sole laboratory in NSW offering a test should be for interstate patients. The proportion should be even

higher for sole laboratories operating in the regions which have a smaller proportion of the Australian population. The fact that the average proportion of assays performed on interstate samples for single region tests was only 5% highlights that patients residing outside the region in which the test is done had limited access to that test.

7.4 Overseas testing

The 426 samples sent for testing overseas accounted for only 0.3% of assays done during 2006. However, many laboratories were unable to provide accurate data on the number of samples sent or the types of test that had been requested in that year. It is likely that the number of samples sent overseas for testing was higher than reported.

A total of 58 types of tests were specified as being requested from an overseas laboratory. At least one Australian laboratory was offering sequencing of the gene in 2006 for 25 (43%) of these types of test. However, the Survey did not collect sufficient data to determine whether the requirements of the requesting laboratory could have been addressed by the Australian laboratory offering sequencing of the gene.

Sequencing was not offered in Australia for 33 of these types of test (57%). The tests involved were ABCA12, ABCA4, ABCB11, ATP7B, BEST1, CDKN2A, COL3A1, COL4A5, DKC1, EDAR, ENG, EYA1, FH, FRG1, IKBKG, MAA, MYH3, NPHS2, PAX3, PAX6, PHEX, PHOX2B, PKD1, PRSS1, REN, RPS19, SALL4, SOX10, SPINK1, TBX5, TEK, TFAP2B, and ZEB2.

8 Characteristics of tests

8.1 Test accreditation

Of the 52 laboratories which provided data for the Survey,

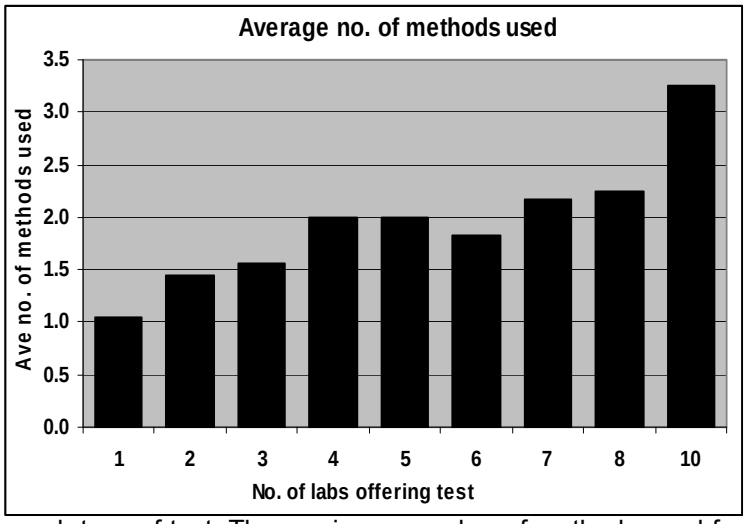
- 28 (54%) only offered accredited tests,
- 6 (11%) only offered non-accredited tests, and
- 18 (35%) provided a mix of both accredited and non-accredited tests.

Of the 437 types of test offered,

- 365 tests (83%) were only offered as accredited tests. This included 208 tests that were only available through one laboratory nationally. The number of laboratories offering these accredited types of test ranged from 1 to 10 per test.
- 16 tests (4%) were only offered as non-accredited tests. 12 of these tests were only available through one laboratory nationally, and the remaining four were available through two laboratories.
- 33 tests (8%) were offered as both accredited and non-accredited tests. All of these tests were offered by two or more laboratories nationally.
- Accreditation was not specified for 23 types of test (5%) from two laboratories. Each test was available through only one laboratory nationally.

The data regarding the number of laboratories in a region offering accredited or non-accredited testing of a gene are tabulated in Section 15.2.1.

8.2 Test methods



Laboratories were asked to provide a simple categorisation of the method used for each type of test in each patient group (see Section 2.8). Analysis for common mutations, or for mutations already identified in the family ie "family" testing, is technically much simpler than diagnostic testing which may involve exhaustive examination of an entire gene sequence.

Methodological data were provided for diagnostic testing of 365 different genes (data in Sections 15.1.9 and 15.2.3). On average, 1.3 different methods were used by various laboratories for each type of test. The maximum number of methods used for a test was four. The Figure demonstrates a direct relationship between the number of laboratories offering a type of test and the average number of methods used for the test (data in Section 15.1.9).

8.3 Test sensitivity

The Survey did not seek to document the analytical performance of each test. However, the Survey did note, in general terms, the method used for testing in different clinical settings and the sensitivity of testing expected by the laboratory. This estimate of sensitivity was the laboratory's expectation of the sensitivity based on experience and publications; it was not the result of a quality assessment.

For family testing, the mutation present in the family has already been defined. The sensitivity of a test for that mutation is, by definition, 100% and so analytical performance for family testing is not addressed.

8.3.1 SCREENING

Screening typically involves testing for a limited number of common mutations using a specific assay. The were 59 reports from laboratories regarding 45 different types of screening test using specific assays. There were a further eight reports of screening tests using more complex methods such as sequencing (with and without MLPA) and mutation screening; these reports may represent a misunderstanding in the definition of the method categories and they are not considered further in this report.

The Table lists the method, the number of types of test based on that method, the number of reports, the range of expected sensitivities, and number of discordant sensitivities i.e. expected sensitivity for the same method and type of test varying by >20% in different laboratories.

Method	*No. of tests*	*No. of reports*	*Sens. range*	*No. of discordant reports*
Specific assays	45	59	<20% to >94%	3

8.3.2 DIAGNOSTIC TESTING

Diagnostic testing involves searching for a mutation, and laboratories typically use different methods that have different sensitivities. Test sensitivity data in a diagnostic setting were reported for 295 tests.

The Table lists the method, the number of types of test based on that method, the number of reports, the range of expected sensitivities, and number of discordant sensitivities i.e. expected sensitivity for the same method and type of test varying by >20% in different laboratories.

Method	*No. of tests*	*No. of reports*	*Sens. range*	*No. of discordant reports*
Mutation screening	48	48	60% to >94%	nil
Sequencing	146	174	20% to >94%	4
Sequencing plus MLPA	51	101	60% to >94%	2
Specific assays	108	177	<20% to >94%	2
FISH	25	47	60% to >94%	1

8.3.3 SOMATIC TESTING

Analysis for somatic mutations can utilise a number of different methodologies. Sensitivity data were reported for 66 tests.

The Table lists the method, the number of types of test based on that method, the number of reports, the range of expected sensitivities, and number of discordant sensitivities i.e. expected sensitivity for the same method and type of test varying by >20% in different laboratories.

Method	*No. of tests*	*No. of reports*	*Sens range*	*No. of discordant reports*
Mutation screening	2	2	80% to >94%	nil
Sequencing	3	3	80% to >94%	nil
Specific assays	29	66	40% to >94%	8
FISH	32	51	>94%	nil

9 Frequency of abnormal test results

The frequency with which a test is abnormal will reflect both the sensitivity of the method used and the selection of patients for the test. A high frequency of abnormal results might suggest that the laboratory's method is highly sensitive, or that clinical indications for testing are too stringent with patients having less characteristic features of the disorder being denied appropriate testing. Conversely, a low-frequency of abnormal results might suggest that the test methodology has a low sensitivity or that the clinical indications for testing are too loose. In other words, the frequency with which a test is abnormal reflects characteristics of both the clinicians requesting the test and the laboratory providing the test.

Laboratories reported the frequency of abnormal results for 220 diagnostic tests. These frequencies were converted into five categories ("ABN categories" in the Table) of 20% increments i.e. 0-19%, 20-39%, 40-59%, 60-79% and 80-100%. The concordance of the 142 reports describing tests offered by two or more laboratories is summarised below. (The 78 reports describing tests offered by a single laboratory were, by definition, concordant in the reported frequency of abnormal results and are not included).

		No. of ABN categories reported by labs			
No. of labs doing test	*No. of tests*	*1*	*2*	*3*	*4*
2	63	55	8		
3	26	22	4		
4	14	7	6	1	
5	15	7	6	2	
6	11	3	7	1	
7	5	2	2	1	
8	4	2	1		1

10	4	2	1	1

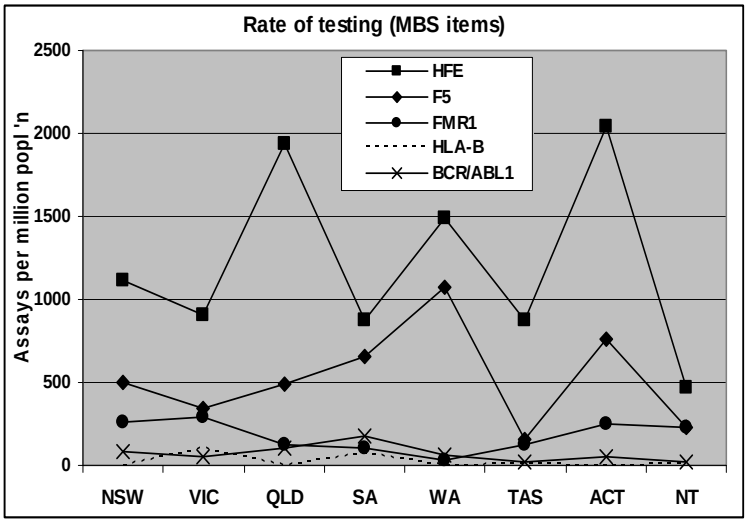
For the majority of tests, the reported frequencies of abnormal results were usually concordant irrespective of the number of laboratories providing the test. For example, of the 26 types of test provided by three laboratories, the three laboratories reported the same frequency of abnormal results for 22 tests. But for four of these tests, the reported frequencies of abnormal results were distributed over two categories. As the number of laboratories doing a type of test increased, the reported frequencies of abnormal results were scattered over an increasing number of categories.

The comparatively wide ranges of test sensitivities (Section 8.2) and frequencies of abnormal results raises the possibility that, for many tests, there is a lack of consistency in patient and method selection.

10 Rates of testing by regions

10.1 Rate of testing for Medicare-rebated tests

The five tests funded by Medicare (BCR/ABL1, F5, FMR1, HFE, and HLA-B) accounted for 25% of the molecular genetic assays performed in Australia in 2006. These tests were available at no financial disadvantage to people in all regions. Nonetheless, there was marked variation in the rate at which these tests were done during 2006.



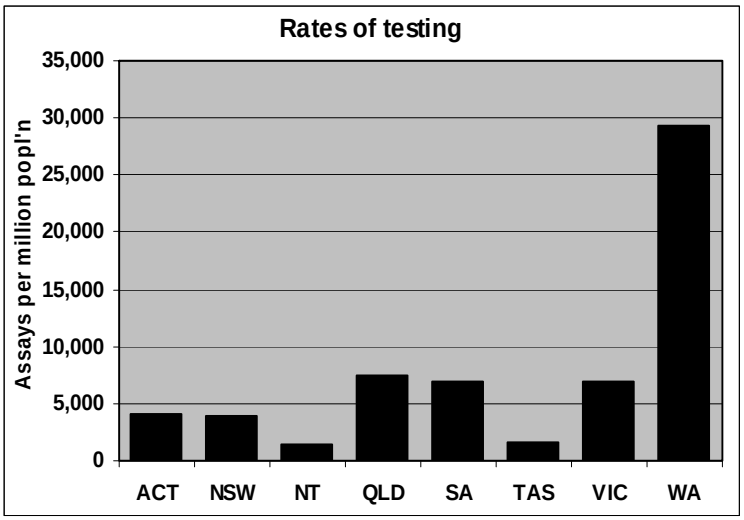
The Figure documents the number of assays per million population for each of these five types of test (data in Section 15.1.10). The rate of testing varied from 4-fold to more than 10-fold across regions for the different tests. These variations were not consistent by region i.e. there was no single region which had the highest or lowest rates for all tests, and the pooled rate of tests by region was less marked and varied 3.3-fold (Section 4)

10.2 Pooled rate of testing for non-Medicare tests

The Medicare data are provided on the basis of the patient's region of residence, thus simplifying comparisons of the rate of testing between regions. The bulk of the non-Medicare tests done during 2006 were for intrastate samples, but a proportion of the assays related to interstate samples. Hence some adjustment must be made for these interstate samples in calculating the rate of testing for non-Medicare tests. The laboratories indicated the number of assays for each test that related to interstate samples, but did not specify the regions from which the samples had been received. For the purpose of calculating rates of testing by region, these interstate samples were allocated to other regions on the basis of the relative populations in those regions. It is recognized that this is unlikely to represent accurately the source of interstate samples referred to a laboratory, but there were no other data to direct how these interstate samples should be apportioned.

The rate of testing in each region for each gene is tabulated in Section 15.2.4. The rate of testing varied widely, with the greatest difference between regions being, on average, 257-fold. The median for the greatest difference between regions was 11-fold. These figures exclude tests for which the rate of testing in any region was zero (and hence the largest ratio of differences between regions was infinite).

The Figure summarises the pooled data for all tests for each region (data in Section 15.1.11). The rate of molecular genetic testing varied by over more than an order of magnitude between regions, with the lowest rate being 21 times less than the highest rate. This difference is 2- to 4-fold greater than regional differences in testing rates noted for Medicare-rebated types of genetic test (Section 10.1).



The high level of testing in WA reflects tissue typing of multiple genes by genetic rather than immunological means; there was only a low volume of such testing by genetic means in other regions. If WA is excluded from the analysis, the greatest difference in pooled testing rates in the remaining regions was 5-fold.

This analysis underestimates the actual difference as it was assumed that the number of interstate assays is distributed uniformly across all regions (other than the one in which the testing laboratory is located). This assumption is essentially the same as the question being asked: is the rate of testing the same in different regions? It would be more accurate to assess the rate of intrastate versus interstate testing for each gene (see Section 10.3). Nonetheless, despite this assumption reducing the variation in estimated testing rates in different regions, substantial differences in rates of testing were identified.

10.3 Rate of testing for each non-Medicare test

The rate of testing for an individual genetic test is very low compared with the population size, and can be modelled using the Poisson distribution. For each test, the rate of testing for intrastate samples was taken as the mean rate; if testing was provided in multiple regions, the mean rate was calculated from the pooled assay volumes and the pooled populations. The rate of testing for interstate samples was estimated on the basis of the number of interstate assays (Section 15.2.1) and the population in regions that did not have a laboratory providing that test (Section 15.2.1). This assessment could not be completed on 108 tests because they had no intrastate assays reported, precluding calculation of the mean rate of testing. Medicare-rebated and overseas tests were excluded.

The rate of intrastate versus interstate testing was compared for each of the remaining 328 tests using the Poisson distribution. 74% of these had a p-value of 0.0001 or lower[7], indicating a significant reduction in the rate of testing provided to patients living outside the region in which the testing was done (data in Section 15.2.5). This confirms that the marked variation in testing rates identified in the pooled data reflects marked variations in the rates of testing for most genes.

[7] A p-value of 0.0001 represents a conservative threshold for determining statistical significance in this setting. There were 328 comparisons, and so the usual threshold of p = 0.05 should be reduced by a factor of 328 i.e. p = 0.00015, to make allowance for these comparisons.

11 Molecular genetic testing in 2007

The data for this Section is in Section 15.2.1.

11.1 Types of test

During 2007, the same number of types of test were nominally provided as in 2006. But, as discussed in Section 5.1, this overestimates the real level of test diversity available during year. The challenge lies in determining whether a lack of testing in one year represented low demand for a rare test, or lack of provision of the test.

There were four types of test for which no assays were performed during 2006 and more than 10 assays were performed in 2007; this could represent an increase in test availability of 0.9%. Conversely, there were seven types of test for which more than 10 assays were performed in 2006 and no assays were performed in 2007; this could represent a decline in test availability of 1.6%. The net change would be a **loss** of three types of test (0.7%). But the threshold of 10 assays is a stringent one for drawing this conclusion. During 2006, 28% of all types of test had assay volumes of between one and 10. If a threshold of 2 assays is used, there were 44 new types of test introduced in 2007 and 11 ceased, a net **increase** of 7.6%.

11.2 Assay volumes

During 2007, the volume of all Medicare-rebated testing increased by 7% compared with 2006. As shown in the Table, the assay volume increased by 3-9% for most Groups. The exception was Medicare-rebated molecular genetic testing which increased by 90%. In 2007, this accounted for 0.12% of all Medicare-rebated tests, up by 0.05% since 2006.

Group	*YR2006*	*YR2007*	*% increase In 2007*
P1Haem	13,842,185	14,393,977	4%
P2 Chem	29,385,064	32,134,761	9%
P3 Micro	8,844,425	9,356,662	6%
P4 Immun	2,153,199	2,330,808	8%
P5 Histol	2,218,777	2,300,512	4%
P6 CYTOL	1,905,817	1,953,894	3%
P7 MolGen	**41,497**	**78,806**	**90%**
P7 CytoGen	45,646	47,556	4%
P8 REPRO	463,066	482,441	4%
P9 SIMPLE	665,979	643,791	-3%
Total	**59,565,655**	**63,723,208**	**7%**

The assay volumes for the five tests rebated by Medicare are shown below. The most dramatic proportional increases related to tests that were only introduced in 2006, reflecting the uptake of a new test rather than an expansion of an existing pattern of testing.

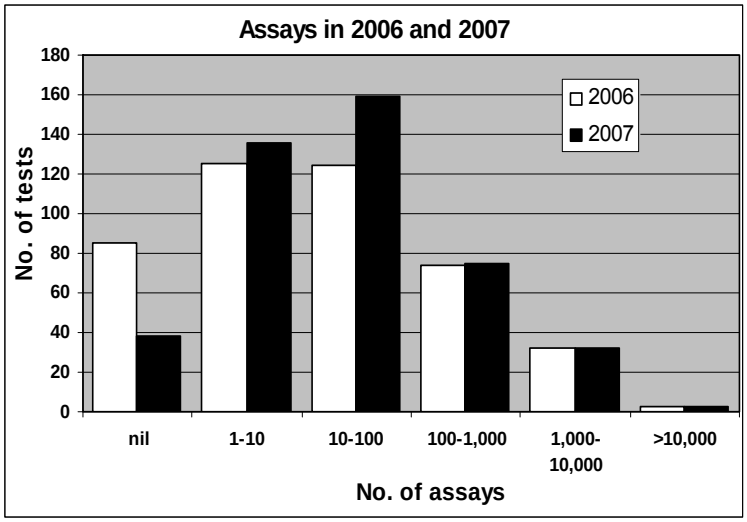
Test	*YR2006*	*YR2007*	*% increase*	*YR introduced*
BCR/ABL1 [MBS]	1,631	3,833	235%	2006
F5 [MBS]	10,338	19,548	189%	2006
FMR1 [MBS]	4,083	4,506	110%	2003
HFE [MBS]	24,767	49,020	198%	2006
HLA-B [MBS]	678	1,899	280%	2006 & 2007

In addition to the Medicare tests, a further 117,342 molecular genetic tests were provided by laboratories using non-Medicare funding. This was a reduction in assay volume of 2,012 (1.7%)

compared with 2006. In 2007, the molecular genetic tests rebated by Medicare accounted for 40% of the molecular genetic tests provided overall, an increase of 14% since 2006. The total number of molecular genetic assays (196,148) is equivalent to 0.3% of all pathology investigations rebated by Medicare during the year; this was an increase of 0.1% since 2006.

11.3 Assays per type of test

This Figure documents the shift in the assay volume per type of test from 2006 to 2007 for each test (data in Section 15.1.4). Medicare-funded testing has been included and counted separately e.g. “F5 [MBS]” and “F5” are counted as separate tests.



11.4 Assays in different patient groups

The Table documents the number of assays performed in different patient groups during 2007. As noted previously, the descriptors for the Medicare items for HFE and FMR1 do not differentiate between diagnostic and family testing. The assay volume for Medicare-rebated HFE almost doubled in 2007, and this accounts for the increased proportion of tests provided to the "unknown" group.

	Assays (n) 2006	*Assays (n) 2007*	*Change from 2006*
Screening	**64,547**	**60,960**	**-6%**
Diagnostic	**45,437**	**55,902**	**+23%**
Somatic	**13,092**	**16,094**	**+23%**
Family	**7,614**	**7,775**	**+2%**
Pharmacogenetic	**470**	**944**	**+101%**
Supplementary	**281**	**362**	**+29%**
Unknown	**29,411**	**54,110**	**+84%**
Total	**160,851**	**196,147**	**+22%**

12 Acknowledgements

This Survey would not have been possible without the financial support of the *Quality Use of Pathology Program* of the Australian Department of Health & Ageing. The encouragement, advice, and support of colleagues in the RCPA and HGSA is gratefully acknowledged. Special thanks to Leslie Burnett, Jacqueline Armstrong, and Mike Ralston.

The greatest “thank you” must go to the many laboratory colleagues who provided the data that underpins this Survey. As per the principle of confidentiality regarding which laboratories provided data, they must remain nameless. But there were many hours - many “after hours” – spent in gathering the data. Thank you.

13 Reference

Gulley ML, Braziel RM, Halling KC, Hsi ED, Kant JA, Nikiforova MN et al (2007). Clinical laboratory reports in molecular pathology. Arch Pathol Lab Med 131:852-863.

14 Appendices

14.1 Letter of invitation

Dear Dr XXXXXX,

Re RCPA/HGSA survey of genetic testing in Australia;

With the continuing "explosion" in genetic knowledge, there is an increasing gap between the genetic testing that medical laboratories would like to provide and the resources that are available. The provision of such resources will probably require support by national and regional authorities, as well as the private sector, and there are a number of models of service provision that could be developed.

Discussions about the demand for and provision of genetic testing in Australia have been hampered by the lack of data regarding the current level of demand and supply. The Royal College of Pathologists of Australasia (RCPA) has received funding from the Australian Department of Health & Ageing for a survey to fill this gap. The project is being overseen by both the RCPA and the Human Genetics Society of Australasia (HGSA). The outcomes of this survey will be a report that is in the public domain and a website (hosted by the RCPA and HGSA) that lists providers of each genetic test.

The principle aim of this survey and the resulting report is to provide data to inform research and policy developments rather than to present recommendations. Hence the report will not provide commentary about the testing currently provided in Australia or make recommendations about future policy in relation to genetic testing. Of course, the report may be the basis for subsequent documents developed by professional organisations, health services etc that include critiques and recommendations regarding genetic testing.

The success of such a survey depends on the cooperation of laboratory services providing genetic testing. *We would be grateful for your support*.

We are seeking to document the utilisation of DNA- and RNA-based testing of human genes for medical purposes during 2006. Please note that the following types of testing lie outside the scope of this survey:

- Medicare-funded tests (Medicare-funded activity will be included in the final report using the Medicare website as the source of these data)
- Research testing done for a specific project (but testing done by a research lab as either *ex gratia* or fee-for-service should be included)
- Testing for non-medical purposes e.g. paternity testing
- Testing of non-human genomes e.g. microbiological testing
- Tests that are not DNA-based and provide surrogate data about human genes e.g. serum cholesterol level as a surrogate marker for heritable mutations in the LDLR gene (causing familial hypercholesterolaemia).

This survey is being sent to all private, public sector, and academic laboratories that provide medical genetic testing in Australia. We have attempted to be inclusive, contacting people listed on HGSA or NATA websites. If you are aware of a medical testing laboratory that has not been approached, would you please let me (Graeme Suthers) know. If you are part of a multi-lab organisation, you may wish to discuss this survey with colleagues to ensure that data are collected without duplication or omissions.

At the outset, we are acutely aware that there may be sensitivities regarding the collection and use of this information. As a matter of principle, we would argue that gathering such information is essential if we are to develop equitable access to genetic testing for the community. But we also recognize the need to be careful about how such information is used. We propose the following precautions:

- The raw data, as captured on the attached survey, will remain confidential. These data will not be made available to the oversight committee for this project, the RCPA, HGSA, or any State or Federal Government Department. The raw data will be handled by only one person (Graeme Suthers), and a confidentiality agreement is attached.
- In the report, the data will be summarised and may presented on a state-by-state basis. The data will not be presented in such a way that within-State or between-laboratory comparisons can be made. We recognize that this may *de facto* identify the only laboratory in a particular State which provides a particular service. Nonetheless, as State funding is a major consideration in the provision of genetic testing, we have to consider regional comparisons.

The purpose of collecting this information is to answer the following questions regarding genetic testing in Australia in 2006:

- What variety of genetic tests (for both heritable and somatic variants) was offered across Australia?
- Were these tests provided as NATA-accredited assays?
- To what extent did laboratories provide testing for their own States versus test samples from interstate?
- What type and volume of genetic tests were sent to overseas laboratories?
- What was the volume of testing in different patient groups i.e. affected, unaffected/predictive, screening, somatic tests?
- What is the anticipated change in test volume in each patient group in 2007? Longer term extrapolations are probably not warranted - and certainly not very accurate!
- Was the utilisation of these tests (tests per 100,000 people) similar across the different States?
- What was the estimated sensitivity of the test methods used?
- What proportion of tests in each patient group was abnormal? The goal is not to compare testing "success" rates (we do not collect sufficient data to draw any conclusions); we only want to document what differences in mutation detection rate exist.

One of the outcomes for this project will be an joint RCPA/HGSA website that would allow your lab to offer testing for specific genes. This will be an updated version of the list currently provided by the HGSA. One of the survey questions (for each gene) is whether you would like to have your lab and this test listed on the website. Prices would not be listed.

I have attached a confidentiality agreement (green), a printed copy of the survey form (blue; the real one will be an EXCEL file), and a draft version of the report (mauve). These documents indicate how the data will be collected and presented.

If you agree to provide these data, I would be grateful if you would sign and return the attached Confidentiality Agreement. I will co-sign and post the original back to you. **Please include the name and email address of the person to whom I can send the EXCEL file for recording your data**. We hope to complete data collection by the end of January.

Thank you in anticipation,

Yours sincerely,

Graeme Suthers.

14.2 Confidentiality Agreement

GENETIC TESTING ACROSS AUSTRALIA SURVEY:
CONFIDENTIALITY AGREEMENT

I, **Graeme Suthers**, being the Survey Coordinator of this RCPA/QUPP project, hereby agree and confirm that

1. All information provided by any participating laboratory to me in my capacity as Coordinator of this project will remain confidential at all times, by which is meant:
   - individual laboratory data will only be seen by myself
   - the Project Committee will only have access to consolidated state-based data.

2. During the conduct of the Project or at any time afterwards I will not disclose or use, in any manner whatsoever, the confidential information supplied to me by any laboratory except for the express purpose for which it was supplied, and only then in consolidated form.

3. Confidential information includes data supplied by the participating laboratory and knowledge gained subsequent to collation and analysis of this data. It does not include information which at the time of its receipt was in the public domain.

4. All laboratories will be issued with a unique identification code. All data should be submitted by participating laboratories using this code. The only person with access to the identification of the individual laboratories will be the Survey Coordinator.

5. All reasonable steps will be taken to maintain the confidentiality and security of all identifiable information.

6. The raw data supplied by participating laboratories will be destroyed once it has been collated in the final report of the survey. The collated data, as represented in the final report, will remain the property of the Royal College of Pathologists of Australasia.

7. Collated and de-identified data collected in the course of this project may be presented at relevant scientific meetings and may be published as part of the proceedings of the RCPA/QUPP Genetic Services Project.

Agreed on behalf of the participating laboratory: _____

Signed: _____ **Date:** _____

Name: _____

Please provide the name and email address of the person who should receive the electronic survey form:

NAME_____**EMAIL**_____

Please return form by fax 08-8161 7984, *or*
by post Dr G Suthers, Familial Cancer Unit, Women's & Children's Hospital, North Adelaide SA 5006.
I will sign below and return the original to you.

Signed: _____ **Date:** _____
Graeme Suthers

14.3 Guidelines for completing questionnaire

GENETIC TESTING ACROSS AUSTRALIA SURVEY:
Guideline for completing the survey

Thank you for agreeing to complete this survey. We very much appreciate your involvement and recognise that this is yet another demand on your time. The RCPA and HGSA are keen to provide a useful and relevant report of genetic testing across Australia, and we have strived to keep the survey instrument straightforward.

The purpose of collecting this information is to answer the following questions regarding genetic testing in Australia in 2006:

1. What variety of genetic tests (for both heritable and somatic variants) was offered across Australia?
2. Were these tests provided as NATA-accredited assays?
3. To what extent did laboratories provide testing for their own States versus test samples from interstate?
4. What type and volume of genetic tests were sent to overseas laboratories?
5. What was the volume of testing in different patient groups i.e. affected, unaffected/predictive, screening, somatic tests?
6. What is the anticipated change in test volume in each patient group in 2007? Longer term extrapolations are probably not very accurate!
7. Was the utilisation of these tests (tests per 100,000 people) similar in different patient groups across the different States?
8. What was the estimated sensitivity of the test methods used?
9. What proportion of tests in each patient group was abnormal? The goal is not to compare testing

"success" rates (we do not collect sufficient data to draw any conclusions); we only want to document what differences in mutation detection rate exist.

We are acutely aware that there may be sensitivities regarding the collection and use of this information. The raw data (as provided by you) will only be seen by the survey coordinator (Graeme Suthers); it will not be made available to the RCPA/HGSA oversight committee, other professional bodies, or any government department. A confidentiality agreement to this effect has been signed.

We are seeking data from all labs that have provided medical testing – including those in the public, private, and academic sectors. If you are working in a multi-lab organisation, please check with colleagues to ensure that you are neither duplicating nor omitting survey data.

Scope of survey

We are seeking information abut the following tests:

- DNA- and RNA-based testing of human genes for medical purposes.
- Testing for both heritable and non-heritable (somatic) genetic variants.
- The samples being tested were collected within Australia.
- The samples were tested during the 2006 calendar year.
- The testing was either performed in an Australian laboratory, or sent from an Australian laboratory to an overseas laboratory (including New Zealand).
- Testing was performed with non-Medicare funds. Please do not include tests that were funded by Medicare (we will obtain these data from Medicare Australia). But please do include data about any tests that are listed on the Medicare schedule but that were performed using non-Medicare funds.

The survey excludes

- Testing done using Medicare funds.
- Medical testing of non-human genes (e.g. microbial genetic testing).
- Non-medical testing of human genes (e.g. paternity testing).
- Testing done principally for research purposes in relation to a specific project. But if the testing reflected *ex gratia* provision of service (usually due to the lack of testing by other means) or was fee-for-service, please include these data in the survey.
- Testing performed on samples received from overseas (including New Zealand).

In general, a test of a specific gene (or locus) for a patient is counted only once on the spreadsheet.

- Multiple assays of the one gene (using different methods) from the one patient counts as one test.
- Multiple assays of one or more loci in multiple patients (e.g. as a segregation or linkage study in a family) count as one “test” per family member (please also specify in the comment field how many loci are usually used for the analysis).
- Samples that were sent interstate for testing will be counted by the receiving (i.e. testing) lab. You do not need to record this activity.
- Samples that were sent overseas for testing need to be counted by the sending lab (i.e. you), please (further details below).
- If a sample is both tested in Australia and subsequently sent interstate or overseas for further testing, it will be counted twice: once by you (when you test), and again either by you (if sent overseas) and by the testing lab (if sent interstate).

Survey instrument

The survey is in the form of an Excel spreadsheet. Your laboratory has been provided with a unique identification code, and this is listed as the worksheet name in the attached file. The first few rows of the spreadsheet contain sample data (these cannot be modified).

The options available are available in a drop-down list (to simplify the collection and analysis of data); please provide comments if you need to clarify an entry. You can copy and paste from row to row if required.

Gene

- Please list each gene on a separate row. As described below, it may be necessary to list a gene on multiple rows to distinguish between testing for different purposes e.g. diagnostic, predictive, screening etc or with different methods.
- Please use conventional gene names (ideally using HUGO nomenclature).
- If a single test involves assays of multiple genes e.g. a panel of pharmacogenetic variants, this can be listed as a single test. Please provide details of the genes involved in the comment field.

Add to website

- Do you want to offer testing in this gene through a RCPA/HGSA website? Your response is not binding, and laboratories will be contacted to confirm details before any listing is placed on the website. At this stage, we do not anticipate that the website will list test prices.

NATA accredited

- If your laboratory was not accredited for genetic testing by NATA during 2006, the answer to this question is "no".
- If your lab was accredited during 2006, was this test (as performed in this patient group) included within the scope of practice of your laboratory's NATA accreditation? Answer "yes" if the test was included in the Medical Testing Assessment Questionnaire (Schedule 1) provided to NATA prior to your last assessment, or if the lab had completed a validation protocol as required by NATA.
- If your lab was accredited during 2006 but the test was either outside your accredited scope of practice or not validated, please answer "no".

Reason for testing

- For each gene being tested, please create a separate row for diagnostic testing, predictive testing, screening, and testing for somatic variants.
    - "Diagnostic" refers to testing of an affected patient (of any age, including prenatal) to determine the genetic basis for their disease.
    - "Predictive" refers to testing of an unaffected person (of any age, including prenatal) who is at increased risk of carrying the mutation on the basis of family history. This will usually refer to testing for a mutation already identified in the family, and includes predictive/presymptomatioc testing, and carrier testing. In certain circumstances, it would also refer to testing an unaffected person for a mutation that has not yet been characterised in the family. For example, testing the child of a person who died of Huntington disease would qualify as predictive testing, even though the precise mutation (size of expansion) responsible for the parent's illness had not been documented.
    - "Screening" refers to testing an unaffected person who is not recognised as being at increase risk of carrying a mutation. This includes neonatal screening for cystic fibrosis, or screening a patient for pharmacogenetic variants prior to commencing drug therapy.
    - "Somatic" refers to testing for non-heritable variants, typically in cancer tissue.
    - "Unknown" refers to testing for unknown purposes. We would like to keep this to a minimum, please!

Method

- Please specify the method used for testing the gene in this group of patients (differentiating between diagnostic, predictive, screening, and somatic).
    - "specific assay/s" refers to any assay for a specific mutation or epimutation. This would include testing for one or more specific sequence variants, or sizing a specific allele (as in testing for a triplet repeat mutation), or testing for a specific abnormality of methylation (as in Prader-Willi syndrome), and screening for deletions in DMD. Please note that any methodology could be included, such as PCR or Southern for allele sizing. The key distinction is that this test focussed on a specific mutation or class of mutations in a gene, and did not search for other mutations in the gene.
    - "mutation screen" refers to screening for unspecified variants by an method such as DHPLC, SSCP, DGGE, PTT etc that is recognised as potentially missing sequence variants.
    - "sequencing only" refers to sequencing of the coding regions of the gene (and adjacent intronic regions) to identify unspecified variants.
    - "sequencing plus dosage" refers to sequencing of the gene plus assays for duplication/deletion of exons (or larger re-arrangements) to detect unspecified variants. Dosage assays include MLPA, QPCR, FISH, and Southern blots for rearrangements.
    - "Southern" refers to a Southern or Northern blot study that is not done as a specific assay or as part of a sequencing/dosage study.
    - "segregation study" refers to a study based on the segregation of genotypes or haplotypes within a pedigree.
    - "sent overseas" refers to samples sent overseas (including New Zealand) by your laboratory. Please create a separate row for such samples (new row for each test type sent overseas). If you first test a sample in your lab, and then send the troublesome cases overseas for further analysis, please list your analysis in one row, and the overseas referrals in another row. For example, *your lab may have tested the CFTR gene from 50 children with cystic fibrosis for 20 common*

mutations. This activity would be included as diagnostic testing of the CFTR gene in 50 samples using "specific assay/s". The lab then sent 10 samples overseas for further analysis; this activity would be coded as diagnostic testing of the CFTR in 10 samples using "sent overseas". Hence these 10 samples will be counted twice on the survey (first tested by you, then tested overseas). **Please do NOT include samples sent <u>interstate</u> for initial or subsequent testing; these data will be collected elsewhere** (see below).

- "see comment" can be used if you cannot fir your methods into the above categories.

- If the lab uses multiple methods, please select the "weakest point" in the analysis. For example, if SSCP is used to screen some exons, and sequencing is used for other exons, the greater potential for missing a variant (i.e. lower sensitivity) lies with the SSCP component of the test. Please code the test method which has the lowest sensitivity.
- If a lab uses different protocols depending on the source of samples, it may be necessary to create two rows. Consider the situation in which a lab tests local samples and some interstate samples using a comprehensive "sequencing plus dosage" protocol. The record of this activity could be included in the one row i.e. diagnostic testing using sequencing plus dosage, with X samples coming from the State and Y samples being received from interstate. But in addition, the lab also provides MLPA/dosage studies alone for some interstate sample in which the sequencing component is completed in the State of origin. The record of this activity should be recorded in a new row i.e. diagnostic testing using specific assay/s, with zero samples coming from within the State and Y samples being sent from interstate.

STATE in 2006

- Please list the volume of tests done on in the patient groups described above (i.e. diagnostic, predictive, screening, somatic, unknown) from your State during the 2006 calendar year.
- If the test was offered but no testing was done, please list the test volume as "0".

STATE in 2007

- Please list the change in test volume for this patient group that you have seen during 2007. Please express this as a percentage of the 2006 experience e.g. "200%" means that the test volume is approximately double what it was in 2006; "50%" means that the test volume has halved in 2007.

Other State in 2006

- Please list the volume of tests done on in the patient groups described above (i.e. diagnostic, predictive, screening, somatic, unknown) from <u>other</u> Australian States or Territories during the 2006 calendar year.
- If the test has been offered but no testing was done, please list the test volume as "0".
- Do not include tests done for overseas patients (including New Zealand).

Other State in 2007

- Please list the change in test volume for the interstate patient group that you have seen during 2007. Please express this as a percentage of the 2006 experience e.g. "200%" means that the test volume is approximately double what it was in 2006; "50%" means that the test volume has halved in 2007.

Sensitivity

- What do you estimate the sensitivity of your assay for this gene (in this patient group) was during 2006? i.e. what proportion of all clinically relevant mutations in this gene would be detected by your lab?
- For recessive disorders, the sensitivity is the proportion of people with the disease in whom <u>both</u> mutations can be identified. Testing for a recessive disorder may identify one mutation, but not both. This does not clarify the diagnosis (the patient may simply be a carrier, and have a different disease).
- For predictive testing, the sensitivity is, by definition, 100%.

ABN%

- Of the tests of this gene among this patient group during 2006, what proportion had an abnormal result?
- For recessive disorders, this refers to the proportion of people in whom two mutations (homozygous or compound heterozygote) were identified.

Comment

Please feel free to clarify any issue. Additional documents also welcome.

DataCheck
If there is something in every column (excluding the comment field), you get a "thank you". If not, there is a red reminder that some data are missing!

*

Thanks again for collating these data. We anticipate that these data will be used by various groups – professional bodies, health services etc – to seek improvements in the resourcing of genetic testing in Australia. Thank you for your contribution to this endeavour.

Graeme Suthers

15 Tabulated data

Electronic copies of this Report (as a WORD file) is available at www.rcpa.edu.au.

15.1 Data for figures

15.1.1 POPULATION DATA

Regional population data were obtained from the Australian Bureau of Statistics in April 2008 at http://www.censusdata.abs.gov.au/ABSNavigation/prenav/PopularAreas?ReadForm&prenavtabname=Popular%20Locations&type=popular&&navmapdisplayed=true&javascript=true&textversion=false&collection=Census&period=2006&producttype=QuickStats&method=&productlabel=&breadcrumb=PL&topic=&.

Figures were not available for 2007, and the estimates below were derived as a linear extrapolation of data from preceding years (1998-2006).

YEAR	NSW	VIC	QLD	SA	WA	TAS	ACT	NT	National
YR2006	6,549,177	4,932,422	3,904,532	1,514,337	1,959,088	476,481	324,034	192,898	19,852,969
YR2007	*6,786,468*	*5,062,771*	*4,047,875*	*1,542,650*	*2,023,650*	*483,325*	*330,487*	*200,927*	*20,478,153*

15.1.2 MEDICAL TESTING IN 2006

Number of assays in each MBS group in 2006 expressed as tests per million population. The data were accessed from http://www.medicareaustralia.gov.au/statistics/dyn_mbs/forms/mbsgtab4.shtml in April 2008. Non-test items such as the patient episode initiation item were excluded.

MBS Group	NSW	VIC	QLD	SA	WA	TAS	ACT	NT	National
P1 Haem	763,014	699,586	675,499	710,226	580,163	616,516	539,107	395,826	697,235
P2 Chem	1,590,051	1,500,998	1,375,589	1,498,013	1,387,480	1,235,791	1,348,297	956,645	1,480,134
P3 Micro	473,540	407,494	457,009	425,430	445,407	340,706	421,113	690,297	445,496
P4 Immuno	135,525	89,879	101,742	93,255	101,116	71,014	120,571	66,501	108,457
P5 Histol	118,534	86,213	155,999	90,844	95,738	91,078	89,290	55,345	111,760
P6 Cytol	102,761	62,114	113,319	108,677	108,072	108,754	112,973	99,866	95,997
P8 Repro	26,469	21,625	22,028	19,900	23,895	13,096	31,083	19,606	23,325
P9 Simple	36,950	30,502	37,254	28,927	30,990	27,050	12,921	33,608	33,546
P7 MolGen	1,882	1,651	2,579	1,837	2,557	1,182	3,048	919	2,018
P7 CytoGen	2,241	1,808	2,915	1,557	2,664	1,105	1,978	1,638	2,219

15.1.3 LABORATORIES OFFERING TYPE OF TEST

No. of labs offering test	No. of tests
1	243
2	94

3	35
4	20
5	16
6	13
7	7
8	5
10	4
Total	437

15.1.4 ASSAYS PER TYPE OF TEST

No. of assays per test	No. of tests (2006)	No. of tests (2007)
nil	85	38
1-10	125	136
10-100	124	159
100-1,000	74	75
1,000-10,000	32	32
>10,000	3	3
Total	443	443

15.1.5 TYPES OF TEST PER LABORATORY

No. of tests offered	No. of labs
<10	24
10-19	12
20-29	11
30-39	1
50-59	1
60-69	2
100-109	1
Total	52

15.1.6 ASSAYS PER LABORATORY

No. of assays per lab	No. of labs
<10	3
10-99	6
100-999	28
1,000-9,999	14
>10,000	1
Total	52

15.1.7 TYPES OF TEST PER REGION

No. of regions offering test	No. of tests
1	247
2	96
3	39
4	27
5	27
6	1
Total	437

15.1.8 ASSAYS FOR INTERSTATE PATIENTS

No. of regions offering test	Average % of assays per test done on interstate samples
1	69%
2	27%
3	23%
4	13%
5	12%
6	3%

15.1.9 AVERAGE NUMBER OF METHODS USED FOR A TEST

No. of labs offering the test	Average no. of methods used for test
1	1.1
2	1.4
3	1.6
4	2.0
5	2.0
6	1.8
7	2.2
8	2.3
10	3.3

15.1.10 RATE OF TESTING FOR MEDICARE ITEMS

Number of assays for the test indicated in 2006 expressed as tests per million population. The data were accessed from http://www.medicareaustralia.gov.au/statistics/dyn_mbs/forms/mbsgtab4.shtml in April 2008.

Test	NSW	VIC	QLD	SA	WA	TAS	ACT	NT	National
F5	500.1	340.0	493.0	661.0	1067.8	159.5	762.3	233.3	520.7
HFE	1116.6	907.3	1940.3	871.0	1486.9	879.4	2043.0	466.6	1247.5
HLA-B	2.3	108.9	1.0	73.3	0.5	14.7	3.1	15.6	34.2
FMR1	265.5	293.2	125.8	100.4	33.7	130.1	253.1	233.3	205.7
BCR/ABL1	88.1	49.5	100.4	175.7	62.3	18.9	49.4	25.9	82.2

15.1.11 POOLED RATE OF TESTING FOR NON-MEDICARE ITEMS

Number of assays for all non-Medicare tests in 2006 expressed as tests per million population for each region.

ACT	NSW	NT	QLD	SA	TAS	VIC	WA	National
4,044	3,946	1,394	7,471	6,999	1,642	6,909	29,389	8019

15.2 Main data tables

In some of the tables, columns are coloured grey to aid reading the tables over multiple pages.

15.2.1 NUMBER OF ASSAYS BY TYPE OF TEST, REGION, AND PATIENT GROUP.

Type of test HGNC test name, or non-standard name (indicated by #).
Region State or Territory.
Patient Group type of patients tested.
Region 06 number of assays done in the region on patient samples from the region in 2006.
Other 06 number of assays done in the region on patient samples from outside the region in 2006.
TOTAL 06 total number of assays done in the region in 2006.
Region 07 number of assays done in the region on patient samples from the region in 2007.
Other 07 number of assays done in the region on patient samples from outside the region in 2007.
TOTAL 07 total number of assays done in the region in 2007.

Type of test	Region	Patient Group	Region 06	Other 06	TOTAL 06	Region 07	Other 07	TOTAL 07
ABCC8	QLD	diagnostic	0	4	**4**	1	3	**4**
ABCD1	SA	diagnostic	2	6	**8**	0	2	**2**
ABCD1	SA	family	6	16	**22**	0	9	**9**
ABL1	WA	somatic	8	0	**8**	64	0	**64**
ACADM	NSW	diagnostic	2	0	**2**	0	2	**2**
ACADM	NSW	family	1	0	**1**	0	3	**3**
ACADM	QLD	screening	30	0	**30**	30	0	**30**
ACADM	QLD	supplementary	11	0	**11**	11	0	**11**
ACADM	SA	diagnostic	3	33	**36**	0	0	**0**
ACADM	SA	screening	52	0	**52**	52	0	**52**
ACADM	VIC	diagnostic	6	0	**6**	6	2	**8**
ACADM	VIC	family	6	0	**6**	3	0	**3**
ACADM	WA	diagnostic	10	0	**10**	12	0	**12**
ACADS	QLD	diagnostic	0	0	**0**	1	1	**2**
ACADVL	WA	family	0	0	**0**	0	0	**0**
ACTA1	WA	diagnostic	2	28	**30**	3	12	**15**
ADA	WA	diagnostic	0	0	**0**	0	0	**0**
ADSL	QLD	diagnostic	0	0	**0**	1	2	**3**
AFF2	SA	diagnostic	6	4	**10**	7	4	**10**
ALDOB	NSW	diagnostic	2	6	**8**	2	4	**6**

ALDOB	QLD	diagnostic	1	0	**1**	2	0	**2**
ALDOB	WA	diagnostic	2	0	**2**	6	0	**6**
ALK	SA	somatic	5	0	**5**	4	1	**5**
ALK	WA	somatic	15	0	**15**	3	1	**4**
ALPL	WA	diagnostic	0	0	**0**	0	0	**0**
APC	NSW	diagnostic	28	0	**28**	34	1	**35**
APC	NSW	family	24	0	**24**	19	1	**20**
APC	QLD	diagnostic	13	0	**13**	17	0	**17**
APC	QLD	family	34	0	**34**	40	0	**40**
APC	SA	diagnostic	23	8	**31**	13	12	**25**
APC	SA	family	11	3	**14**	22	1	**24**
APC	SA	supplementary	7	0	**7**	7	1	**8**
APC	VIC	diagnostic	90	36	**126**	67	12	**79**
APC	VIC	family	47	18	**65**	38	26	**64**
APC	WA	diagnostic	13	0	**13**	10	0	**10**
APC	WA	family	0	0	**0**	6	0	**6**
APOB	NSW	diagnostic	0	0	**0**	13	1	**14**
APOB	SA	diagnostic	6	0	**6**	7	0	**7**
APOB	WA	diagnostic	20	0	**20**	20	1	**21**
APOE	NSW	screening	60	0	**60**	48	0	**48**
APOE	QLD	diagnostic	187	0	**187**	220	0	**220**
APOE	QLD	screening	193	0	**193**	212	0	**212**
APOE	SA	diagnostic	360	0	**360**	400	0	**400**
APOE	VIC	diagnostic	1	0	**1**	2	1	**3**
APOE	VIC	screening	461	0	**461**	453	1207	**1660**
APOE	WA	diagnostic	100	0	**100**	146	1	**147**
APP	VIC	diagnostic	31	10	**41**	9	12	**21**
AR	NSW	diagnostic	24	4	**28**	15	0	**15**
AR	NSW	family	2	0	**2**	0	0	**0**
AR	QLD	diagnostic	32	10	**42**	32	9	**41**
AR	SA	diagnostic	16	0	**16**	12	1	**13**
AR	SA	family	4	0	**4**	0	2	**2**
AR	VIC	diagnostic	30	3	**33**	60	6	**66**
AR	WA	diagnostic	16	0	**16**	6	0	**6**
ARSA	SA	diagnostic	2	1	**3**	3	0	**3**

ARSA	SA	family	3	5	**8**	2	1	**3**
ARSE	WA	diagnostic	0	0	**0**	0	0	**0**
ARVCF	SA	diagnostic	2	0	**2**	29	0	**29**
ARX	SA	diagnostic	19	2	**21**	32	4	**36**
ARX	WA	diagnostic	1	0	**1**	1	0	**1**
ASPA	NSW	diagnostic	1	0	**1**	0	0	**0**
ASPA	NSW	family	7	0	**7**	1	0	**1**
ASPA	NSW	screening	362	0	**362**	326	0	**326**
ASPA	SA	diagnostic	0	0	**0**	3	0	**3**
ASPA	VIC	screening	79	0	**79**	104	1	**105**
ATM	NSW	somatic	7	0	**7**	42	1	**43**
ATM	SA	somatic	35	0	**35**	37	1	**38**
ATM	WA	diagnostic	1	0	**1**	2	0	**2**
ATM	WA	family	0	0	**0**	1	0	**1**
ATN1	NSW	diagnostic	2	0	**2**	5	0	**5**
ATN1	NSW	family	0	0	**0**	0	0	**0**
ATN1	QLD	diagnostic	126	0	**126**	130	0	**130**
ATN1	SA	diagnostic	47	0	**47**	34	1	**35**
ATN1	VIC	diagnostic	3	1	**4**	6	1	**7**
ATN1	VIC	family	4	0	**4**	0	0	**0**
ATN1	WA	diagnostic	3	1	**4**	4	2	**6**
ATP13A2	VIC	diagnostic	0	0	**0**	0	2	**2**
ATP1A2	QLD	diagnostic	0	0	**0**	3	1	**4**
ATP7A	VIC	family	12	0	**12**	0	0	**0**
ATRX	WA	diagnostic	0	0	**0**	2	0	**2**
ATXN1	NSW	diagnostic	51	3	**54**	46	5	**51**
ATXN1	NSW	family	1	0	**1**	0	0	**0**
ATXN1	QLD	diagnostic	126	0	**126**	130	0	**130**
ATXN1	SA	diagnostic	47	0	**47**	34	1	**35**
ATXN1	VIC	diagnostic	107	1	**108**	59	6	**65**
ATXN1	VIC	family	9	0	**9**	3	1	**4**
ATXN1	WA	diagnostic	53	0	**53**	57	0	**57**
ATXN2	NSW	diagnostic	58	3	**61**	41	6	**47**
ATXN2	NSW	family	0	0	**0**	2	0	**2**
ATXN2	QLD	diagnostic	126	0	**126**	130	0	**130**

ATXN2	SA	diagnostic	47	0	**47**	34	1	**35**
ATXN2	VIC	diagnostic	107	1	**108**	59	7	**66**
ATXN2	VIC	family	7	0	**7**	1	1	**2**
ATXN2	WA	diagnostic	53	0	**53**	55	0	**55**
ATXN3	NSW	diagnostic	54	3	**57**	49	5	**54**
ATXN3	NSW	family	0	0	**0**	2	0	**2**
ATXN3	QLD	diagnostic	126	0	**126**	130	0	**130**
ATXN3	SA	diagnostic	47	0	**47**	34	1	**35**
ATXN3	VIC	diagnostic	107	1	**108**	59	7	**66**
ATXN3	VIC	family	7	0	**7**	1	1	**2**
ATXN3	WA	diagnostic	57	0	**57**	59	0	**59**
ATXN7	NSW	diagnostic	59	3	**62**	50	4	**54**
ATXN7	NSW	family	0	0	**0**	0	0	**0**
ATXN7	QLD	diagnostic	126	0	**126**	130	0	**130**
ATXN7	SA	diagnostic	47	0	**47**	34	1	**35**
ATXN7	VIC	diagnostic	107	1	**108**	59	6	**65**
ATXN7	VIC	family	7	0	**7**	1	0	**1**
ATXN7	WA	diagnostic	55	0	**55**	57	0	**57**
AVP	QLD	diagnostic	0	0	**0**	2	1	**3**
AZF#	NSW	diagnostic	93	0	**93**	93	1	**94**
AZF#	WA	diagnostic	9	0	**9**	74	0	**74**
BCHE	QLD	diagnostic	15	0	**15**	24	0	**24**
BCHE	WA	screening	22	0	**22**	22	1	**23**
BCL2	ACT	somatic	16	0	**16**	16	0	**16**
BCL2	NSW	somatic	13	0	**13**	14	1	**15**
BCL2	QLD	somatic	955	0	**955**	1260	0	**1260**
BCL2	SA	somatic	4	0	**4**	8	1	**9**
BCL2	VIC	somatic	133	0	**133**	113	1	**114**
BCL6	WA	somatic	13	0	**13**	18	1	**19**
BCR/ABL1	NSW	somatic	10	0	**10**	20	1	**21**
BCR/ABL1	QLD	somatic	120	0	**120**	94	0	**94**
BCR/ABL1	SA	somatic	980	1147	**2127**	143	33	**176**
BCR/ABL1	VIC	somatic	483	0	**483**	434	2	**436**
BCR/ABL1	VIC	supplementary	18	0	**18**	17	1	**18**
BCR/ABL1	WA	somatic	12	0	**12**	17	1	**18**

BCR/ABL1 [MBS]	ACT	somatic	16	0	**16**	93	0	**93**
BCR/ABL1 [MBS]	NSW	somatic	577	0	**577**	1455	0	**1455**
BCR/ABL1 [MBS]	NT	somatic	5	0	**5**	7	0	**7**
BCR/ABL1 [MBS]	QLD	somatic	392	0	**392**	746	0	**746**
BCR/ABL1 [MBS]	SA	somatic	266	0	**266**	430	0	**430**
BCR/ABL1 [MBS]	TAS	somatic	9	0	**9**	25	0	**25**
BCR/ABL1 [MBS]	VIC	somatic	244	0	**244**	834	0	**834**
BCR/ABL1 [MBS]	WA	somatic	122	0	**122**	243	0	**243**
BDNF	SA	diagnostic	0	0	**0**	3	0	**3**
BLM	NSW	diagnostic	0	0	**0**	0	0	**0**
BLM	NSW	family	0	0	**0**	0	0	**0**
BLM	NSW	screening	57	0	**57**	52	0	**52**
BLM	VIC	screening	79	0	**79**	104	1	**105**
BMPR1A	NSW	diagnostic	1	1	**2**	1	1	**2**
BMPR1A	NSW	family	2	2	**4**	2	2	**4**
BRAF	NSW	somatic	100	0	**100**	200	0	**200**
BRAF	SA	somatic	1	0	**1**	22	1	**23**
BRAF	VIC	somatic	7	2	**9**	31	2	**33**
BRAF	WA	diagnostic	0	0	**0**	6	0	**6**
BRCA1	NSW	diagnostic	321	1	**322**	387	6	**393**
BRCA1	NSW	family	72	3	**75**	113	4	**117**
BRCA1	QLD	diagnostic	73	0	**73**	81	0	**81**
BRCA1	QLD	family	28	0	**28**	54	0	**54**
BRCA1	SA	diagnostic	98	14	**112**	86	24	**110**
BRCA1	SA	family	47	3	**50**	73	15	**88**
BRCA1	VIC	diagnostic	197	155	**352**	246	223	**469**
BRCA1	VIC	family	56	8	**64**	84	24	**108**
BRCA1	VIC	supplementary	5	6	**11**	13	51	**64**
BRCA1	WA	diagnostic	103	0	**103**	128	0	**128**
BRCA1	WA	family	3	0	**3**	42	0	**42**
BRCA2	NSW	diagnostic	304	1	**305**	387	6	**393**
BRCA2	NSW	family	77	5	**82**	99	2	**101**
BRCA2	QLD	diagnostic	73	0	**73**	81	0	**81**
BRCA2	QLD	family	28	0	**28**	54	0	**54**
BRCA2	SA	diagnostic	101	13	**114**	88	26	**114**

BRCA2	SA	family	32	5	**37**	57	3	**60**
BRCA2	VIC	diagnostic	191	163	**354**	236	228	**464**
BRCA2	VIC	family	45	14	**59**	68	29	**96**
BRCA2	VIC	supplementary	5	6	**11**	13	51	**64**
BRCA2	WA	diagnostic	101	0	**101**	125	0	**125**
BRCA2	WA	family	0	0	**0**	48	0	**48**
BTK	WA	diagnostic	2	0	**2**	2	0	**2**
CACNA1A	NSW	diagnostic	56	3	**59**	49	5	**54**
CACNA1A	NSW	family	0	0	**0**	0	0	**0**
CACNA1A	QLD	diagnostic	128	22	**150**	137	29	**166**
CACNA1A	SA	diagnostic	47	0	**47**	34	1	**35**
CACNA1A	SA	family	5	0	**5**	2	1	**3**
CACNA1A	VIC	diagnostic	107	1	**108**	59	6	**65**
CACNA1A	VIC	family	7	0	**7**	1	0	**1**
CACNA1A	WA	diagnostic	57	0	**57**	59	0	**59**
CACNA1S	QLD	diagnostic	6	0	**6**	2	0	**2**
CACNA1S	WA	diagnostic	6	2	**8**	9	6	**15**
CASR	QLD	diagnostic	9	0	**9**	19	0	**19**
CASR	WA	diagnostic	11	2	**13**	4	0	**4**
CAV3	WA	diagnostic	1	0	**1**	2	2	**4**
CBFB	NSW	somatic	6	0	**6**	3	1	**4**
CBFB	SA	somatic	6	0	**6**	4	1	**5**
CBFB/MYH11	VIC	somatic	7	0	**7**	1	1	**2**
CBFB/MYH11	WA	somatic	13	0	**13**	15	0	**15**
CCND1	QLD	somatic	554	0	**554**	787	0	**787**
CCND1	VIC	somatic	20	0	**20**	23	1	**24**
CCND1	WA	somatic	3	0	**3**	5	0	**5**
CCND1/IGHG1	NSW	somatic	27	0	**27**	30	1	**31**
CCND1/IGHG1	SA	somatic	79	0	**79**	17	1	**18**
CCND1/IGHG1	WA	somatic	5	0	**5**	10	1	**11**
CCR5	WA	screening	44	0	**44**	44	1	**45**
CD109	VIC	screening	1140	20	**1160**	1140	20	**1160**
CD109	WA	diagnostic	30	0	**30**	47	0	**47**
CD177	VIC	screening	10	0	**10**	10	1	**11**
CD2	WA	diagnostic	0	0	**0**	0	0	**0**

CD40LG	WA	diagnostic	0	0	**0**	2	0	**2**
CDC2L1	SA	diagnostic	17	0	**17**	7	0	**7**
CDC2L1	WA	diagnostic	6	0	**6**	5	1	**6**
CDC45L	SA	diagnostic	2	0	**2**	29	0	**29**
CDC73	WA	diagnostic	1	0	**1**	1	0	**1**
CDH1	NSW	diagnostic	6	2	**8**	6	3	**9**
CDH1	NSW	family	0	3	**3**	3	9	**12**
CDH1	VIC	diagnostic	0	0	**0**	2	0	**2**
CDH1	VIC	family	0	0	**0**	40	0	**40**
CDKN1B	SA	diagnostic	3	0	**3**	6	0	**6**
CDKN1C	VIC	diagnostic	0	0	**0**	1	10	**11**
CDKN2A	NSW	family	1	2	**3**	0	2	**2**
CFTR	ACT	diagnostic	10	0	**10**	10	0	**10**
CFTR	ACT	screening	21	0	**21**	21	0	**21**
CFTR	NSW	diagnostic	87	3	**90**	19	4	**23**
CFTR	NSW	family	138	0	**138**	103	5	**108**
CFTR	NSW	screening	4434	0	**4434**	3288	7	**3295**
CFTR	NSW	supplementary	172	0	**172**	132	1	**133**
CFTR	QLD	screening	2280	0	**2280**	2280	0	**2280**
CFTR	SA	diagnostic	94	1	**95**	94	1	**95**
CFTR	SA	family	236	3	**239**	236	2	**238**
CFTR	SA	screening	279	31	**310**	279	30	**309**
CFTR	VIC	diagnostic	294	25	**319**	267	28	**295**
CFTR	VIC	family	1146	130	**1276**	1339	139	**1478**
CFTR	VIC	screening	1016	130	**1146**	1199	140	**1339**
CFTR	WA	diagnostic	566	0	**566**	701	0	**701**
CFTR	WA	family	0	0	**0**	76	0	**76**
CHD7	WA	diagnostic	1	0	**1**	4	0	**4**
CHD7	WA	family	0	0	**0**	1	0	**1**
Chimerism#	NSW	somatic	1	0	**1**	2	1	**3**
Chimerism#	SA	somatic	12	0	**12**	36	1	**37**
Chimerism#	VIC	diagnostic	0	0	**0**	1	3	**4**
Chimerism#	WA	somatic	33	0	**33**	56	0	**56**
CLCN1	WA	diagnostic	6	15	**21**	11	11	**22**
CLDN5	SA	diagnostic	2	0	**2**	29	0	**29**

CLIP2	SA	diagnostic	2	0	**2**	29	0	**29**
CLN3	SA	diagnostic	2	13	**15**	0	10	**10**
CLN3	SA	family	0	11	**11**	0	1	**1**
CLN3	SA	screening	0	1	**1**	0	0	**0**
CLTCL1	SA	diagnostic	2	0	**2**	29	0	**29**
COL2A1	VIC	family	3	0	**3**	0	0	**0**
COL3A1	NSW	family	4	0	**4**	2	1	**3**
COL7A1	QLD	diagnostic	9	0	**9**	12	0	**12**
CPOX	NSW	diagnostic	0	0	**0**	4	1	**5**
CPS1	VIC	family	0	0	**0**	3	0	**3**
CPS1	WA	diagnostic	0	0	**0**	1	0	**1**
CREBBP	SA	diagnostic	0	0	**0**	3	0	**3**
CREBBP	WA	diagnostic	1	7	**8**	2	8	**10**
CRK	SA	diagnostic	3	0	**3**	0	0	**0**
CRK	WA	diagnostic	3	0	**3**	6	1	**7**
CTBP1	SA	diagnostic	0	0	**0**	3	0	**3**
CTLA4	VIC	screening	20	0	**20**	20	1	**21**
CTNND2	SA	diagnostic	0	0	**0**	3	0	**3**
CTNS	SA	diagnostic	1	2	**3**	0	0	**0**
CTNS	SA	family	2	2	**4**	0	3	**3**
CTSB	SA	diagnostic	0	0	**0**	3	0	**3**
CYBB	WA	diagnostic	1	0	**1**	0	0	**0**
CYP21A2	QLD	diagnostic	3	10	**13**	1	20	**21**
CYP21A2	QLD	family	8	16	**24**	3	38	**41**
CYP21A2	QLD	screening	0	3	**3**	3	7	**10**
CYP21A2	WA	diagnostic	12	0	**12**	19	0	**19**
CYP21A2	WA	family	0	0	**0**	8	0	**8**
CYP2C19	VIC	pharmacogenetic	100	40	**140**	200	80	**280**
CYP2C9	SA	screening	0	0	**0**	1	1	**2**
CYP2C9	VIC	pharmacogenetic	60	35	**95**	120	70	**190**
CYP2D6	VIC	pharmacogenetic	100	40	**140**	200	80	**280**
D13S319#	NSW	somatic	0	0	**0**	60	1	**61**
D19S545#	WA	somatic	32	0	**32**	64	1	**65**
D19S851#	WA	somatic	32	0	**32**	64	1	**65**
D4Z4#	NSW	diagnostic	50	34	**84**	33	58	**91**

D4Z4#	NSW	family	8	14	**22**	6	6	**12**
D4Z4#	WA	diagnostic	32	1	**33**	23	2	**25**
D5S721#	SA	diagnostic	1	0	**1**	3	0	**3**
D7S613#	SA	diagnostic	15	0	**15**	13	0	**13**
D7S613#	WA	diagnostic	15	0	**15**	30	1	**31**
DARC	SA	diagnostic	8	0	**8**	5	0	**5**
DARC	VIC	diagnostic	10	0	**10**	0	0	**0**
DARC	VIC	family	25	1	**26**	17	0	**17**
DARC	WA	diagnostic	5	0	**5**	5	0	**5**
DCX	WA	diagnostic	2	11	**13**	2	14	**16**
DDIT3	NSW	somatic	0	0	**0**	3	1	**4**
DDIT3	SA	somatic	3	0	**3**	1	1	**2**
DGCR	SA	diagnostic	5	0	**5**	117	0	**117**
DGCR2	WA	diagnostic	5	0	**5**	2	1	**3**
DHCR7	SA	diagnostic	0	3	**3**	0	1	**1**
DHCR7	SA	family	0	3	**3**	0	4	**4**
DMD	NSW	diagnostic	21	0	**21**	4	1	**5**
DMD	NSW	unknown	145	70	**215**	132	140	**272**
DMD	QLD	diagnostic	15	0	**15**	16	0	**16**
DMD	SA	diagnostic	11	1	**12**	12	1	**13**
DMD	SA	family	11	1	**12**	10	1	**11**
DMD	VIC	diagnostic	89	2	**91**	59	11	**70**
DMD	VIC	family	132	6	**138**	34	2	**36**
DMD	WA	diagnostic	11	0	**11**	9	0	**9**
DMPK	NSW	diagnostic	62	0	**62**	81	10	**91**
DMPK	NSW	family	18	2	**20**	16	0	**16**
DMPK	QLD	diagnostic	81	0	**81**	63	0	**63**
DMPK	SA	diagnostic	48	3	**51**	39	2	**41**
DMPK	SA	family	17	10	**27**	6	1	**7**
DMPK	VIC	diagnostic	124	24	**148**	113	13	**126**
DMPK	VIC	family	199	9	**208**	79	37	**116**
DMPK	WA	diagnostic	25	0	**25**	24	0	**24**
DYRK1A	SA	diagnostic	0	0	**0**	3	0	**3**
EBP	SA	diagnostic	1	0	**1**	0	0	**0**
EBP	SA	family	1	0	**1**	0	0	**0**

EDA	WA	diagnostic	0	0	**0**	1	0	**1**
EGFR	SA	somatic	12	24	**36**	21	25	**46**
EGFR	VIC	somatic	136	32	**168**	129	33	**162**
EGFR	WA	somatic	32	0	**32**	64	1	**65**
ELN	NSW	diagnostic	1	0	**1**	1	0	**1**
ELN	SA	diagnostic	17	0	**17**	42	0	**42**
ELN	WA	diagnostic	15	0	**15**	30	1	**31**
EMD	VIC	diagnostic	8	9	**17**	2	0	**2**
EMD	VIC	family	20	3	**23**	0	0	**0**
ERBB2	SA	somatic	0	0	**0**	1	1	**2**
ETV6/RUNX1	QLD	somatic	120	0	**120**	94	0	**94**
ETV6/RUNX1	SA	somatic	7	9	**16**	8	9	**17**
ETV6/RUNX1	VIC	somatic	19	0	**19**	38	1	**39**
ETV6/RUNX1	WA	somatic	12	0	**12**	24	1	**25**
EWSR1	NSW	somatic	3	0	**3**	4	1	**5**
EWSR1	SA	somatic	9	0	**9**	13	1	**14**
EWSR1/FLI1	VIC	somatic	3	0	**3**	8	1	**9**
EWSR1/WT1	VIC	somatic	1	0	**1**	3	1	**4**
EXT1	SA	diagnostic	0	0	**0**	3	0	**3**
EXT1	SA	somatic	5	0	**5**	2	0	**2**
F11	SA	diagnostic	19	0	**19**	9	0	**9**
F12	NSW	diagnostic	0	200	**200**	0	0	**0**
F2	QLD	diagnostic	912	0	**912**	820	0	**820**
F5	QLD	diagnostic	912	0	**912**	820	0	**820**
F5 [MBS]	ACT	diagnostic	223	0	**223**	406	0	**406**
F5 [MBS]	ACT	family	24	0	**24**	57	0	**57**
F5 [MBS]	NSW	diagnostic	2839	0	**2839**	6274	0	**6274**
F5 [MBS]	NSW	family	436	0	**436**	875	0	**875**
F5 [MBS]	NT	diagnostic	35	0	**35**	66	0	**66**
F5 [MBS]	NT	family	10	0	**10**	5	0	**5**
F5 [MBS]	QLD	diagnostic	1568	0	**1568**	1799	0	**1799**
F5 [MBS]	QLD	family	357	0	**357**	655	0	**655**
F5 [MBS]	SA	diagnostic	933	0	**933**	1948	0	**1948**
F5 [MBS]	SA	family	68	0	**68**	126	0	**126**
F5 [MBS]	TAS	diagnostic	59	0	**59**	140	0	**140**

F5 [MBS]	TAS	family	17	0	**17**	45	0	**45**
F5 [MBS]	VIC	diagnostic	1503	0	**1503**	3128	0	**3128**
F5 [MBS]	VIC	family	174	0	**174**	423	0	**423**
F5 [MBS]	WA	diagnostic	1894	0	**1894**	3293	0	**3293**
F5 [MBS]	WA	family	198	0	**198**	308	0	**308**
F8	NSW	diagnostic	3	0	**3**	3	1	**4**
F8	NSW	family	12	0	**12**	9	1	**10**
F8	QLD	diagnostic	46	0	**46**	18	0	**18**
F8	SA	diagnostic	25	22	**47**	13	11	**24**
F8	SA	family	24	16	**40**	27	8	**35**
F8	VIC	diagnostic	44	0	**44**	91	0	**91**
F8	WA	diagnostic	8	0	**8**	10	0	**10**
F9	NSW	diagnostic	0	0	**0**	3	1	**4**
F9	SA	diagnostic	9	4	**13**	5	4	**9**
F9	SA	family	1	2	**3**	2	14	**16**
FAH	QLD	diagnostic	0	0	**0**	1	1	**2**
FANCC	NSW	diagnostic	0	0	**0**	0	0	**0**
FANCC	NSW	family	0	0	**0**	2	0	**2**
FANCC	NSW	screening	366	0	**366**	333	0	**333**
FANCC	VIC	screening	79	0	**79**	104	1	**105**
FBN1	NSW	diagnostic	14	7	**21**	0	7	**7**
FBN1	NSW	family	17	15	**32**	0	8	**8**
FBN1	WA	diagnostic	0	0	**0**	0	0	**0**
FCGR3A	VIC	screening	10	0	**10**	10	1	**11**
FDFT1	SA	diagnostic	0	0	**0**	3	0	**3**
FGFR1	NSW	diagnostic	50	56	**106**	46	41	**86**
FGFR1	NSW	family	6	0	**6**	0	0	**0**
FGFR1	VIC	diagnostic	28	2	**30**	28	8	**36**
FGFR1	VIC	family	15	0	**15**	2	0	**2**
FGFR1	WA	diagnostic	8	0	**8**	22	0	**22**
FGFR2	NSW	diagnostic	50	56	**106**	46	41	**86**
FGFR2	NSW	family	5	0	**5**	0	0	**0**
FGFR2	VIC	diagnostic	28	2	**30**	28	8	**36**
FGFR2	VIC	family	15	0	**15**	2	0	**2**
FGFR2	WA	diagnostic	8	0	**8**	23	0	**23**

FGFR2	WA	family	0	0	**0**	2	0	**2**
FGFR3	NSW	diagnostic	72	57	**129**	58	61	**119**
FGFR3	NSW	family	6	0	**6**	0	10	**10**
FGFR3	QLD	diagnostic	22	0	**22**	11	0	**11**
FGFR3	SA	diagnostic	0	0	**0**	3	0	**3**
FGFR3	SA	somatic	30	0	**30**	3	1	**4**
FGFR3	VIC	diagnostic	28	2	**30**	28	8	**36**
FGFR3	VIC	family	14	0	**14**	2	0	**2**
FGFR3	WA	diagnostic	11	0	**11**	26	0	**26**
FGFR3	WA	family	0	0	**0**	5	0	**5**
FGFRL1	SA	diagnostic	0	0	**0**	3	0	**3**
FIP1L1	NSW	somatic	3	0	**3**	6	1	**7**
FIP1L1/PDGFRA	NSW	somatic	38	52	**90**	59	56	**115**
FIP1L1/PDGFRA	VIC	somatic	16	0	**16**	18	1	**19**
FKRP	WA	diagnostic	0	0	**0**	4	0	**4**
FLCN	NSW	family	6	0	**6**	10	2	**12**
FLCN	WA	family	2	0	**2**	1	0	**1**
FLNA	VIC	family	3	0	**3**	0	0	**0**
FLNA	WA	diagnostic	1	0	**1**	3	0	**3**
FLT3	VIC	somatic	3	0	**3**	3	1	**4**
FLT3	WA	somatic	19	0	**19**	38	0	**38**
FMR1	NSW	screening	44	0	**44**	44	1	**45**
FMR1	SA	diagnostic	519	34	**553**	576	67	**643**
FMR1	SA	family	25	1	**26**	25	2	**27**
FMR1 [MBS]	ACT	unknown	82	0	**82**	101	0	**101**
FMR1 [MBS]	NSW	unknown	1739	0	**1739**	1815	0	**1815**
FMR1 [MBS]	NT	unknown	45	0	**45**	50	0	**50**
FMR1 [MBS]	QLD	unknown	491	0	**491**	531	0	**531**
FMR1 [MBS]	SA	unknown	152	0	**152**	158	0	**158**
FMR1 [MBS]	TAS	unknown	62	0	**62**	66	0	**66**
FMR1 [MBS]	VIC	unknown	1446	0	**1446**	1558	0	**1558**
FMR1 [MBS]	WA	unknown	66	0	**66**	227	0	**227**
FOXC2	WA	diagnostic	1	0	**1**	2	0	**2**
FOXC2	WA	family	0	0	**0**	0	0	**0**
FOXL2	WA	diagnostic	0	0	**0**	2	0	**2**

FRAXA	WA	diagnostic	317	0	**317**	0	0	**0**
FRAXE	WA	diagnostic	317	0	**317**	0	0	**0**
FXN	NSW	diagnostic	19	1	**20**	23	0	**23**
FXN	NSW	family	1	0	**1**	0	0	**0**
FXN	QLD	diagnostic	126	0	**126**	130	0	**130**
FXN	SA	diagnostic	9	0	**9**	5	1	**6**
FXN	VIC	diagnostic	105	9	**114**	65	26	**91**
FXN	VIC	family	161	9	**170**	35	19	**54**
FXN	WA	diagnostic	25	0	**25**	29	0	**29**
FZD9	SA	diagnostic	0	0	**0**	3	0	**3**
G6PC	NSW	diagnostic	0	0	**0**	0	0	**0**
G6PC	NSW	family	0	0	**0**	0	0	**0**
G6PC	NSW	screening	56	0	**56**	4	0	**4**
GABRB3	SA	diagnostic	2	0	**2**	29	0	**29**
GAK	SA	diagnostic	0	0	**0**	3	0	**3**
GALT	QLD	diagnostic	25	0	**25**	31	0	**31**
GALT	SA	diagnostic	1	0	**1**	4	0	**4**
GALT	SA	family	1	0	**1**	4	0	**4**
GALT	WA	diagnostic	3	0	**3**	0	0	**0**
GBA	SA	diagnostic	0	4	**4**	0	3	**3**
GBA	SA	family	0	17	**17**	0	3	**3**
GBA	SA	screening	0	5	**5**	0	4	**4**
GCH1	VIC	diagnostic	0	0	**0**	0	1	**1**
GCK	QLD	diagnostic	1	3	**4**	9	1	**10**
GDAP1	NSW	diagnostic	0	0	**0**	0	0	**0**
GFAP	VIC	family	0	0	**0**	2	0	**2**
GFAP	WA	diagnostic	2	0	**2**	1	0	**1**
GJB1	NSW	diagnostic	13	10	**23**	7	13	**20**
GJB1	NSW	family	0	0	**0**	0	0	**0**
GJB1	WA	diagnostic	12	0	**12**	10	0	**10**
GJB2	NSW	diagnostic	253	0	**253**	7	0	**7**
GJB2	NSW	family	0	0	**0**	2	0	**2**
GJB2	NSW	screening	1	0	**1**	2	0	**2**
GJB2	QLD	diagnostic	91	0	**91**	91	0	**91**
GJB2	SA	diagnostic	81	0	**81**	58	1	**59**

GJB2	SA	family	6	0	**6**	4	1	**5**
GJB2	VIC	diagnostic	121	163	**284**	78	133	**211**
GJB2	VIC	family	349	26	**375**	21	28	**49**
GJB2	WA	diagnostic	42	0	**42**	13	0	**13**
GJB6	NSW	diagnostic	3	0	**3**	7	0	**7**
GJB6	NSW	family	0	0	**0**	2	0	**2**
GJB6	NSW	screening	0	0	**0**	0	0	**0**
GJB6	SA	diagnostic	81	0	**81**	58	1	**59**
GJB6	SA	family	6	0	**6**	4	1	**5**
GJB6	VIC	diagnostic	192	96	**288**	110	138	**248**
GJB6	VIC	family	330	14	**344**	8	8	**16**
GJB6	WA	diagnostic	42	0	**42**	12	0	**12**
GLA	SA	diagnostic	0	9	**9**	0	13	**13**
GLA	SA	family	3	1	**4**	0	13	**13**
GLDC	WA	diagnostic	0	0	**0**	0	0	**0**
GLUD1	QLD	diagnostic	0	0	**0**	1	1	**2**
GNB1	SA	diagnostic	2	0	**2**	29	0	**29**
GP1BA	VIC	screening	1407	20	**1427**	1407	20	**1427**
GP1BA	WA	diagnostic	30	0	**30**	47	0	**47**
GRN	VIC	diagnostic	0	0	**0**	1	1	**2**
H19	VIC	diagnostic	21	39	**60**	15	49	**63**
HADHA	QLD	diagnostic	1	0	**1**	3	1	**4**
HADHA	SA	diagnostic	4	0	**4**	0	2	**2**
HADHA	WA	diagnostic	5	0	**5**	0	0	**0**
HBA1	NSW	diagnostic	118	246	**364**	135	5	**140**
HBA1	SA	diagnostic	349	6	**355**	401	8	**409**
HBA1	SA	family	2	6	**8**	2	4	**6**
HBA1	VIC	diagnostic	774	36	**810**	1393	58	**1451**
HBA1	WA	diagnostic	1117	0	**1117**	1240	1	**1241**
HBA1	WA	family	0	0	**0**	1	0	**1**
HBA2	NSW	diagnostic	118	246	**364**	135	5	**140**
HBA2	SA	diagnostic	349	6	**355**	401	8	**409**
HBA2	SA	family	2	6	**8**	2	4	**6**
HBA2	VIC	diagnostic	774	36	**810**	1393	58	**1451**
HBA2	WA	diagnostic	1117	0	**1117**	1240	1	**1241**

HBB	NSW	diagnostic	114	15	**129**	89	5	**94**
HBB	NSW	family	2	0	**2**	20	1	**21**
HBB	SA	diagnostic	92	9	**101**	83	15	**98**
HBB	SA	family	3	31	**34**	4	8	**12**
HBB	VIC	diagnostic	169	5	**174**	213	13	**226**
HBB	WA	diagnostic	583	0	**583**	639	1	**640**
HBB	WA	family	0	0	**0**	1	0	**1**
HBD	NSW	diagnostic	6	0	**6**	15	1	**16**
HBD	VIC	diagnostic	169	5	**174**	213	13	**226**
HBG1	NSW	diagnostic	12	0	**12**	19	1	**20**
HBG2	NSW	diagnostic	12	0	**12**	19	1	**20**
HEXA	NSW	diagnostic	1	1	**2**	1	1	**2**
HEXA	NSW	family	8	4	**12**	0	0	**0**
HEXA	NSW	screening	456	45	**501**	388	27	**415**
HEXA	VIC	diagnostic	4	2	**6**	0	1	**1**
HEXA	VIC	family	0	0	**0**	0	2	**2**
HEXA	VIC	screening	598	0	**598**	615	1	**616**
HEXB	NSW	diagnostic	0	4	**4**	0	0	**0**
HEXB	NSW	screening	4	4	**8**	0	0	**0**
HFE	NSW	diagnostic	91	0	**91**	112	1	**113**
HFE	QLD	diagnostic	3270	0	**3270**	3467	0	**3467**
HFE	VIC	diagnostic	15	0	**15**	15	1	**16**
HFE [MBS]	ACT	unknown	662	0	**662**	1437	0	**1437**
HFE [MBS]	NSW	unknown	7313	0	**7313**	16746	0	**16746**
HFE [MBS]	NT	unknown	90	0	**90**	164	0	**164**
HFE [MBS]	QLD	unknown	7576	0	**7576**	12721	0	**12721**
HFE [MBS]	SA	unknown	1319	0	**1319**	2382	0	**2382**
HFE [MBS]	TAS	unknown	419	0	**419**	873	0	**873**
HFE [MBS]	VIC	unknown	4475	0	**4475**	9350	0	**9350**
HFE [MBS]	WA	unknown	2913	0	**2913**	5347	0	**5347**
HIC1	SA	diagnostic	2	0	**2**	29	0	**29**
HIPK3	SA	diagnostic	0	0	**0**	3	0	**3**
HIRA	NSW	diagnostic	20	0	**20**	32	1	**33**
HIRA	QLD	diagnostic	12	0	**12**	12	0	**12**
HIRA	SA	diagnostic	39	0	**39**	74	0	**74**

HIRA	WA	diagnostic	118	0	**118**	111	2	**113**
HLA-A	VIC	diagnostic	320	30	**350**	320	30	**350**
HLA-A	WA	screening	4900	0	**4900**	4410	1	**4411**
HLA-B	NSW	screening	249	0	**249**	613	1	**614**
HLA-B	SA	screening	0	0	**0**	0	0	**0**
HLA-B	VIC	diagnostic	200	20	**220**	200	20	**220**
HLA-B	WA	screening	4900	0	**4900**	4410	1	**4411**
HLA-B [MBS]	ACT	diagnostic	1	0	**1**	0	0	**0**
HLA-B [MBS]	ACT	pharmacogenetic	0	0	**0**	0	0	**0**
HLA-B [MBS]	NSW	diagnostic	15	0	**15**	41	0	**41**
HLA-B [MBS]	NSW	pharmacogenetic	0	0	**0**	2	0	**2**
HLA-B [MBS]	NT	diagnostic	3	0	**3**	3	0	**3**
HLA-B [MBS]	NT	pharmacogenetic	0	0	**0**	0	0	**0**
HLA-B [MBS]	QLD	diagnostic	4	0	**4**	8	0	**8**
HLA-B [MBS]	QLD	pharmacogenetic	0	0	**0**	0	0	**0**
HLA-B [MBS]	SA	diagnostic	111	0	**111**	134	0	**134**
HLA-B [MBS]	SA	pharmacogenetic	0	0	**0**	2	0	**2**
HLA-B [MBS]	TAS	diagnostic	7	0	**7**	6	0	**6**
HLA-B [MBS]	TAS	pharmacogenetic	0	0	**0**	0	0	**0**
HLA-B [MBS]	VIC	diagnostic	537	0	**537**	1695	0	**1695**
HLA-B [MBS]	VIC	pharmacogenetic	0	0	**0**	0	0	**0**
HLA-B [MBS]	WA	diagnostic	0	0	**0**	8	0	**8**
HLA-B [MBS]	WA	pharmacogenetic	0	0	**0**	0	0	**0**
HLA-C	VIC	diagnostic	80	5	**85**	80	5	**85**
HLA-C	WA	screening	4900	0	**4900**	4410	1	**4411**
HLA-DPB1	VIC	screening	20	0	**20**	20	1	**21**
HLA-DPB1	WA	screening	4900	0	**4900**	4410	1	**4411**
HLA-DQA1	NSW	diagnostic	0	250	**250**	0	0	**0**
HLA-DQA1	VIC	screening	300	10	**310**	300	10	**310**
HLA-DQB1	VIC	screening	300	10	**310**	300	10	**310**
HLA-DQB1	WA	screening	4900	0	**4900**	4410	1	**4411**
HLA-DRB1	NSW	diagnostic	0	250	**250**	0	0	**0**
HLA-DRB1	SA	screening	0	0	**0**	0	0	**0**
HLA-DRB1	VIC	diagnostic	120	20	**140**	120	20	**140**
HLA-DRB1	WA	screening	4900	0	**4900**	4410	1	**4411**

HLA-DRB3	VIC	diagnostic	15	2	**17**	15	2	**17**
HLA-DRB3	WA	screening	4900	0	**4900**	4410	1	**4411**
HLA-DRB4	WA	screening	4900	0	**4900**	4410	1	**4411**
HLA-DRB5	WA	screening	4900	0	**4900**	4410	1	**4411**
HMBS	NSW	diagnostic	0	0	**0**	3	1	**4**
HMGCL	WA	diagnostic	0	0	**0**	0	0	**0**
HMHA1	VIC	screening	5	0	**5**	5	1	**6**
HNF1A	QLD	diagnostic	8	3	**11**	14	1	**15**
HNF4A	QLD	diagnostic	0	1	**1**	5	1	**6**
HPRT1	QLD	diagnostic	0	0	**0**	1	1	**2**
HRAS	WA	diagnostic	0	0	**0**	10	0	**10**
HSD17B4	SA	diagnostic	0	1	**1**	0	0	**0**
HSD17B4	SA	family	0	2	**2**	0	0	**0**
HTT	NSW	diagnostic	84	0	**84**	71	1	**72**
HTT	QLD	unknown	175	0	**175**	168	0	**168**
HTT	SA	diagnostic	33	0	**33**	25	1	**26**
HTT	SA	family	26	0	**26**	24	1	**25**
HTT	VIC	diagnostic	110	11	**121**	144	16	**160**
HTT	VIC	family	316	111	**427**	39	81	**120**
HTT	WA	diagnostic	42	2	**44**	42	1	**43**
HTT	WA	family	31	2	**33**	31	1	**32**
IDS	SA	diagnostic	0	1	**1**	0	1	**1**
IDS	SA	family	1	1	**2**	0	10	**10**
IDUA	SA	family	0	4	**4**	4	16	**20**
IDUA	SA	screening	0	1	**1**	0	0	**0**
IGH@	ACT	somatic	87	2	**89**	100	2	**102**
IGH@	QLD	somatic	955	0	**955**	1260	0	**1260**
IGH@	SA	diagnostic	125	4	**129**	175	4	**179**
IGH@	SA	somatic	35	0	**35**	37	1	**38**
IGH@	VIC	somatic	169	0	**169**	176	1	**177**
IGH@	WA	somatic	41	0	**41**	53	0	**53**
IGH@/BCL2	SA	somatic	60	2	**62**	93	2	**95**
IGH@/BCL2	WA	somatic	31	0	**31**	41	1	**42**
IGH@/CCND1	SA	somatic	21	0	**21**	48	1	**49**
IGH@/CCND1	WA	somatic	7	0	**7**	7	0	**7**

IGH@/MYC	WA	somatic	4	0	**4**	20	1	**21**
IGHG1	NSW	somatic	7	0	**7**	42	1	**43**
IGHG1	WA	somatic	32	0	**32**	64	1	**65**
IGHR	VIC	somatic	18	0	**18**	0	0	**0**
IKBKAP	NSW	diagnostic	0	0	**0**	0	0	**0**
IKBKAP	NSW	family	1	0	**1**	1	0	**1**
IKBKAP	NSW	screening	365	0	**365**	332	0	**332**
IKBKAP	VIC	screening	79	0	**79**	104	1	**105**
IKBKG	SA	diagnostic	18	11	**29**	31	7	**38**
IKBKG	SA	family	2	2	**4**	3	6	**9**
IKBKG	WA	diagnostic	0	0	**0**	0	0	**0**
IL2RG	NSW	diagnostic	1	0	**1**	0	12	**12**
IL2RG	NSW	family	1	0	**1**	0	13	**13**
IL2RG	WA	diagnostic	0	0	**0**	0	0	**0**
IL7R	WA	diagnostic	0	0	**0**	0	0	**0**
ITGA2	VIC	screening	1407	20	**1427**	1407	20	**1427**
ITGA2	WA	diagnostic	30	0	**30**	47	0	**47**
ITGA2B	VIC	diagnostic	367	0	**367**	367	0	**367**
ITGA2B	VIC	screening	1407	20	**1427**	1407	20	**1427**
ITGA2B	WA	diagnostic	30	0	**30**	47	0	**47**
ITGB3	VIC	screening	1784	20	**1804**	1784	21	**1805**
ITGB3	WA	diagnostic	30	0	**30**	47	0	**47**
ITGB4	QLD	diagnostic	11	0	**11**	14	0	**14**
JAG1	NSW	diagnostic	0	0	**0**	0	14	**14**
JAG1	NSW	family	3	6	**9**	0	15	**15**
JAG1	SA	diagnostic	2	0	**2**	29	0	**29**
JAG1	VIC	family	1	0	**1**	0	0	**0**
JAG1	WA	diagnostic	0	0	**0**	0	0	**0**
JAG1	WA	family	1	0	**1**	0	0	**0**
JAK2	NSW	diagnostic	16	0	**16**	117	1	**118**
JAK2	NSW	somatic	351	0	**351**	509	1	**510**
JAK2	QLD	somatic	0	0	**0**	1	0	**1**
JAK2	SA	somatic	252	11	**263**	267	12	**279**
JAK2	VIC	somatic	239	38	**277**	81	38	**119**
JAK2	WA	somatic	70	0	**70**	700	0	**700**

KAL1	SA	diagnostic	7	0	**7**	1	0	**1**
KAL1	WA	diagnostic	2	0	**2**	4	0	**4**
KCNE1	VIC	diagnostic	34	5	**39**	35	13	**48**
KCNE1	VIC	family	9	0	**9**	14	2	**16**
KCNE1	WA	diagnostic	5	0	**5**	2	0	**2**
KCNE2	VIC	diagnostic	34	5	**39**	35	13	**48**
KCNE2	VIC	family	9	0	**9**	14	1	**15**
KCNE2	WA	diagnostic	5	0	**5**	2	0	**2**
KCNH2	VIC	diagnostic	34	5	**39**	35	13	**48**
KCNH2	VIC	family	9	0	**9**	14	2	**16**
KCNH2	WA	diagnostic	5	0	**5**	3	0	**3**
KCNH2	WA	family	0	0	**0**	2	0	**2**
KCNJ11	QLD	diagnostic	0	3	**3**	1	3	**4**
KCNJ2	VIC	diagnostic	34	5	**39**	35	13	**48**
KCNJ2	VIC	family	9	0	**9**	14	2	**16**
KCNJ6	SA	diagnostic	0	0	**0**	3	0	**3**
KCNQ1	VIC	diagnostic	34	5	**39**	35	13	**48**
KCNQ1	VIC	family	9	0	**9**	14	1	**15**
KCNQ1	WA	diagnostic	5	0	**5**	2	0	**2**
KCNQ1OT1	VIC	diagnostic	13	31	**44**	14	26	**40**
KEL	SA	diagnostic	8	0	**8**	5	0	**5**
KEL	VIC	diagnostic	10	0	**10**	0	0	**0**
KEL	VIC	family	25	1	**26**	17	0	**17**
KEL	WA	diagnostic	5	0	**5**	5	0	**5**
KIR3DL1	VIC	screening	30	3	**33**	30	3	**33**
KIT	NSW	family	1	0	**1**	1	1	**2**
KIT	VIC	somatic	109	56	**165**	109	67	**176**
KMS	SA	diagnostic	1	0	**1**	0	0	**0**
KRAS	VIC	somatic	0	0	**0**	0	1	**1**
KRAS	WA	diagnostic	0	0	**0**	8	0	**8**
KRIT1	WA	diagnostic	0	0	**0**	0	0	**0**
KRT10	VIC	family	0	0	**0**	0	7	**7**
KRT14	QLD	diagnostic	18	0	**18**	23	0	**23**
KRT14	QLD	family	1	0	**1**	1	0	**1**
KRT5	QLD	diagnostic	18	0	**18**	23	0	**23**

KRT5	QLD	family	1	0	**1**	1	0	**1**
L1CAM	VIC	family	6	0	**6**	0	0	**0**
L1CAM	WA	diagnostic	3	0	**3**	5	0	**5**
L1CAM	WA	family	1	0	**1**	3	0	**3**
LAMA3	QLD	diagnostic	11	0	**11**	14	0	**14**
LAMB3	QLD	diagnostic	11	0	**11**	14	0	**14**
LAMC2	QLD	diagnostic	11	0	**11**	14	0	**14**
LDLR	NSW	diagnostic	0	0	**0**	52	2	**54**
LETM1	SA	diagnostic	0	0	**0**	3	0	**3**
LIMK1	SA	diagnostic	17	0	**17**	42	0	**42**
LIMK1	WA	diagnostic	15	0	**15**	30	1	**31**
LLGL1	SA	diagnostic	2	0	**2**	29	0	**29**
LMNA	NSW	diagnostic	4	2	**6**	6	3	**9**
LMNA	NSW	family	0	0	**0**	0	0	**0**
LMNA	VIC	diagnostic	19	9	**28**	12	1	**13**
LMNA	VIC	family	31	3	**34**	0	0	**0**
LRRK2	VIC	diagnostic	1	2	**3**	0	2	**2**
MAPT	VIC	diagnostic	1	0	**1**	1	2	**3**
MAPT	WA	diagnostic	6	0	**6**	2	0	**2**
MAT1A	WA	diagnostic	0	0	**0**	3	0	**3**
MBP	WA	diagnostic	0	0	**0**	0	0	**0**
MC2R	QLD	diagnostic	0	3	**3**	4	1	**5**
MC4R	QLD	diagnostic	0	1	**1**	2	1	**3**
MCOLN1	NSW	diagnostic	0	0	**0**	0	0	**0**
MCOLN1	NSW	family	0	0	**0**	0	0	**0**
MCOLN1	NSW	screening	56	0	**56**	50	0	**50**
MECP2	NSW	diagnostic	45	39	**84**	0	16	**16**
MECP2	NSW	family	3	4	**7**	0	17	**17**
MECP2	QLD	diagnostic	0	0	**0**	12	1	**13**
MECP2	WA	diagnostic	64	28	**92**	36	17	**53**
MEFV	NSW	diagnostic	165	98	**263**	0	18	**18**
MEFV	SA	diagnostic	12	2	**14**	12	2	**14**
MEFV	WA	unknown	6	0	**6**	18	1	**19**
MEN1	QLD	diagnostic	21	0	**21**	47	0	**47**
MEN1	SA	diagnostic	2	0	**2**	0	0	**0**

MEN1	VIC	diagnostic	15	5	**20**	30	8	**38**
MEN1	VIC	family	10	5	**15**	20	8	**28**
MEN1	WA	diagnostic	25	0	**25**	8	0	**8**
MEN1	WA	family	0	0	**0**	1	0	**1**
MET	NSW	family	2	0	**2**	3	1	**4**
MET	SA	diagnostic	1	0	**1**	1	1	**2**
MET	SA	family	1	1	**2**	4	3	**7**
MFAP4	SA	diagnostic	2	0	**2**	29	0	**29**
MFN2	NSW	diagnostic	35	6	**41**	46	2	**47**
MFN2	NSW	family	0	0	**0**	0	0	**0**
MFN2	WA	diagnostic	9	0	**9**	15	0	**15**
MGMT	VIC	somatic	0	0	**0**	0	1	**1**
MKRN3	SA	diagnostic	2	0	**2**	29	0	**29**
MLH1	NSW	diagnostic	29	0	**29**	41	1	**42**
MLH1	NSW	family	30	0	**30**	57	1	**58**
MLH1	QLD	diagnostic	15	0	**15**	12	0	**12**
MLH1	QLD	family	45	0	**45**	25	0	**25**
MLH1	SA	diagnostic	18	12	**30**	9	10	**19**
MLH1	SA	family	9	0	**9**	8	3	**11**
MLH1	VIC	diagnostic	64	57	**121**	68	50	**118**
MLH1	VIC	family	60	30	**90**	45	35	**80**
MLH1	WA	diagnostic	106	0	**106**	22	0	**22**
MLH1	WA	family	0	0	**0**	9	0	**9**
MLL	NSW	somatic	7	0	**7**	4	1	**5**
MLL	SA	somatic	19	9	**28**	25	9	**34**
MLL/AFF1	QLD	somatic	120	0	**120**	94	0	**94**
MNX1	WA	diagnostic	0	0	**0**	0	0	**0**
MPZ	NSW	diagnostic	11	3	**14**	0	0	**0**
MPZ	NSW	family	0	0	**0**	0	0	**0**
MPZ	WA	diagnostic	6	0	**6**	5	0	**5**
MSH2	NSW	diagnostic	23	0	**23**	32	1	**33**
MSH2	NSW	family	26	0	**26**	52	1	**53**
MSH2	QLD	diagnostic	12	0	**12**	13	0	**13**
MSH2	QLD	family	69	0	**69**	43	0	**43**
MSH2	SA	diagnostic	12	5	**17**	7	8	**15**

MSH2	SA	family	16	12	**28**	26	10	**36**
MSH2	VIC	diagnostic	51	52	**103**	63	47	**109**
MSH2	VIC	family	60	30	**90**	45	32	**77**
MSH2	WA	diagnostic	106	0	**106**	25	0	**25**
MSH2	WA	family	0	0	**0**	35	0	**35**
MSH6	NSW	diagnostic	4	0	**4**	8	1	**9**
MSH6	NSW	family	11	0	**11**	13	1	**14**
MSH6	SA	diagnostic	3	0	**3**	9	2	**11**
MSH6	SA	family	3	2	**5**	7	5	**12**
MSH6	VIC	diagnostic	52	35	**87**	59	64	**123**
MSH6	VIC	family	42	17	**59**	37	13	**50**
MSH6	WA	diagnostic	105	0	**105**	25	0	**25**
MSH6	WA	family	0	0	**0**	5	0	**5**
MSI#	NSW	somatic	150	0	**150**	225	0	**225**
MSI#	SA	screening	183	0	**183**	9	155	**164**
MSI#	VIC	diagnostic	26	9	**35**	26	2	**28**
MSI#	VIC	somatic	0	0	**0**	0	1	**1**
MSI#	WA	diagnostic	0	0	**0**	7	0	**7**
MSRA	SA	diagnostic	0	0	**0**	3	0	**3**
MT-ATP6	QLD	diagnostic	209	0	**209**	240	0	**240**
MT-ATP6	SA	diagnostic	2	1	**3**	2	3	**5**
MT-ATP6	SA	family	0	0	**0**	1	3	**4**
MT-ATP6	VIC	diagnostic	94	52	**146**	261	52	**313**
MT-ATP6	VIC	family	76	1	**77**	1	0	**1**
MT-ATP6	VIC	supplementary	0	1	**1**	0	3	**3**
MT-CO1	VIC	diagnostic	0	1	**1**	1	3	**4**
MT-CO2	VIC	diagnostic	0	1	**1**	1	3	**4**
MT-CO3	VIC	diagnostic	0	1	**1**	1	3	**4**
MT-CYB	QLD	diagnostic	24	0	**24**	24	0	**24**
MT-deletion#	QLD	diagnostic	84	0	**84**	75	0	**75**
MT-deletion#	VIC	diagnostic	35	2	**37**	58	2	**60**
MTHFR	QLD	diagnostic	132	0	**132**	147	1	**148**
MTHFR	QLD	screening	131	0	**131**	217	0	**217**
MTHFR	VIC	screening	30	0	**30**	30	1	**31**
MTM1	WA	diagnostic	0	3	**3**	0	4	**4**

MT-ND1	QLD	diagnostic	24	0	**24**	24	0	**24**
MT-ND1	SA	diagnostic	10	1	**11**	5	1	**6**
MT-ND1	SA	family	0	0	**0**	1	1	**2**
MT-ND1	VIC	diagnostic	32	9	**41**	46	9	**55**
MT-ND1	VIC	supplementary	0	0	**0**	1	1	**2**
MT-ND4	QLD	diagnostic	233	0	**233**	264	0	**264**
MT-ND4	SA	diagnostic	10	1	**11**	5	1	**6**
MT-ND4	SA	family	0	0	**0**	1	1	**2**
MT-ND4	VIC	diagnostic	32	9	**41**	46	9	**55**
MT-ND4	VIC	supplementary	0	0	**0**	1	1	**2**
MT-ND5	NSW	diagnostic	11	2	**13**	0	26	**26**
MT-ND5	SA	diagnostic	18	2	**20**	17	0	**17**
MT-ND5	SA	family	0	0	**0**	1	1	**2**
MT-ND5	VIC	diagnostic	1	0	**1**	27	2	**29**
MT-ND5	VIC	supplementary	0	0	**0**	1	1	**2**
MT-ND6	SA	diagnostic	10	1	**11**	5	1	**6**
MT-ND6	SA	family	0	0	**0**	1	1	**2**
MT-ND6	VIC	diagnostic	32	9	**41**	46	9	**55**
MT-ND6	VIC	supplementary	0	0	**0**	2	1	**3**
MT-RNR1	SA	diagnostic	2	0	**2**	2	1	**3**
MT-RNR1	SA	family	0	0	**0**	1	1	**2**
MT-RNR1	VIC	diagnostic	0	1	**1**	1	1	**2**
MT-RNR1	WA	diagnostic	1	0	**1**	6	0	**6**
MT-TK	NSW	diagnostic	11	2	**13**	0	26	**26**
MT-TK	QLD	diagnostic	209	0	**209**	240	0	**240**
MT-TK	SA	diagnostic	5	1	**6**	16	3	**19**
MT-TK	SA	family	0	0	**0**	1	1	**2**
MT-TK	VIC	diagnostic	156	56	**212**	305	52	**357**
MT-TK	VIC	family	76	2	**78**	1	0	**1**
MT-TK	VIC	supplementary	0	1	**1**	2	1	**3**
MT-TL1	QLD	diagnostic	209	0	**209**	240	0	**240**
MT-TL1	SA	diagnostic	24	2	**26**	28	2	**30**
MT-TL1	SA	family	0	0	**0**	1	1	**2**
MT-TL1	VIC	diagnostic	96	3	**99**	134	6	**140**
MT-TL1	VIC	supplementary	2	0	**2**	1	1	**2**

MUTYH	NSW	diagnostic	10	4	**14**	40	6	**46**
MUTYH	NSW	family	25	5	**30**	31	8	**39**
MUTYH	SA	diagnostic	164	9	**173**	30	7	**37**
MUTYH	SA	family	0	0	**0**	1	1	**2**
MUTYH	VIC	diagnostic	31	7	**38**	19	8	**27**
MUTYH	VIC	family	47	2	**49**	0	3	**3**
MUTYH	WA	diagnostic	0	0	**0**	17	0	**17**
MUTYH	WA	family	0	0	**0**	3	0	**3**
MYBPC3	VIC	diagnostic	2	0	**2**	15	0	**15**
MYBPC3	VIC	family	2	0	**2**	3	1	**4**
MYC	NSW	somatic	18	0	**18**	25	1	**26**
MYC	SA	somatic	14	0	**14**	14	1	**15**
MYC	WA	somatic	5	0	**5**	11	1	**12**
MYCN	QLD	somatic	43	0	**43**	26	0	**26**
MYCN	VIC	somatic	2	0	**2**	4	1	**5**
MYCN	WA	diagnostic	0	0	**0**	0	0	**0**
MYH7	VIC	diagnostic	0	0	**0**	3	1	**4**
MYH7	VIC	family	3	0	**3**	14	0	**14**
MYH7	WA	diagnostic	0	0	**0**	0	4	**4**
MYOT	WA	diagnostic	0	0	**0**	0	0	**0**
NAGLU	SA	family	0	1	**1**	1	2	**3**
NDN	SA	diagnostic	2	0	**2**	29	0	**29**
NDP	QLD	diagnostic	6	0	**6**	6	0	**6**
NDP	WA	diagnostic	0	0	**0**	0	0	**0**
NF1	WA	diagnostic	13	0	**13**	8	0	**8**
NF1	WA	family	0	0	**0**	0	0	**0**
NF2	SA	diagnostic	0	9	**9**	1	12	**13**
NF2	SA	family	0	2	**2**	1	6	**7**
NF2	VIC	diagnostic	8	0	**8**	37	3	**40**
NF2	VIC	family	8	0	**8**	21	0	**21**
NF2	WA	diagnostic	5	0	**5**	12	0	**12**
NF2	WA	family	0	0	**0**	0	0	**0**
NIPA1	NSW	diagnostic	1	1	**2**	24	10	**34**
NIPA1	NSW	family	0	0	**0**	0	0	**0**
NOD2	ACT	family	0	8	**8**	0	0	**0**

NOD2	VIC	diagnostic	50	0	**50**	13	0	**13**
NOG	WA	diagnostic	0	0	**0**	3	0	**3**
NOTCH3	QLD	diagnostic	1	1	**2**	0	3	**3**
NOTCH3	QLD	family	1	26	**27**	2	30	**32**
NOTCH3	WA	diagnostic	33	43	**76**	38	39	**77**
NPC1	NSW	diagnostic	0	0	**0**	0	0	**0**
NPC1	NSW	family	0	0	**0**	0	0	**0**
NPC1	NSW	screening	56	0	**56**	49	0	**49**
NPC1	SA	diagnostic	0	1	**1**	0	0	**0**
NPC1	SA	family	1	2	**3**	3	4	**7**
NR0B1	WA	diagnostic	0	0	**0**	0	0	**0**
NR4A2	VIC	diagnostic	0	0	**0**	0	1	**1**
NSD1	SA	diagnostic	2	0	**2**	37	0	**37**
NSD1	WA	diagnostic	6	0	**6**	12	0	**12**
OCRL	WA	diagnostic	0	0	**0**	2	0	**2**
OTC	NSW	diagnostic	5	3	**8**	0	19	**19**
OTC	NSW	family	10	2	**12**	0	20	**20**
OTC	QLD	diagnostic	2	0	**2**	1	3	**4**
OTC	SA	family	2	1	**3**	0	0	**0**
OTC	VIC	family	5	0	**5**	3	0	**3**
OTOF	WA	diagnostic	0	0	**0**	9	0	**9**
PABPN1	QLD	diagnostic	21	0	**21**	21	0	**21**
PAFAH1B1	NSW	diagnostic	1	0	**1**	1	0	**1**
PAFAH1B1	SA	diagnostic	5	0	**5**	29	0	**29**
PAFAH1B1	WA	diagnostic	6	0	**6**	6	1	**7**
PAH	QLD	diagnostic	1	0	**1**	14	0	**14**
PALB2	WA	diagnostic	0	0	**0**	0	0	**0**
PANK2	VIC	diagnostic	0	0	**0**	0	1	**1**
PARK2	VIC	diagnostic	5	4	**9**	1	4	**5**
PARK7	VIC	diagnostic	0	1	**1**	0	1	**1**
PAX6	SA	diagnostic	6	0	**6**	5	0	**5**
PAX7/FOXO1A	VIC	somatic	3	0	**3**	5	1	**6**
PDCD6	SA	diagnostic	0	0	**0**	3	0	**3**
PDHA1	VIC	family	1	0	**1**	0	0	**0**
PEX1	SA	family	3	3	**6**	0	2	**2**

PEX1	SA	screening	2	0	**2**	0	0	**0**
PEX7	SA	family	0	3	**3**	0	5	**5**
PINK1	VIC	diagnostic	0	2	**2**	0	2	**2**
PITX2	WA	diagnostic	0	0	**0**	3	0	**3**
PKP2	VIC	family	2	0	**2**	0	0	**0**
PLA2G6	WA	diagnostic	0	0	**0**	2	0	**2**
PLA2G6	WA	family	0	0	**0**	1	0	**1**
PML/RARA	NSW	somatic	17	0	**17**	13	1	**14**
PML/RARA	SA	somatic	6	0	**6**	12	2	**14**
PML/RARA	VIC	somatic	52	0	**52**	25	2	**27**
PML/RARA	WA	somatic	36	0	**36**	56	0	**56**
PMM2	SA	diagnostic	0	4	**4**	0	4	**4**
PMM2	SA	family	0	4	**4**	0	10	**10**
PMP22	NSW	diagnostic	76	42	**118**	57	34	**91**
PMP22	NSW	family	4	1	**5**	1	2	**3**
PMP22	SA	diagnostic	32	0	**32**	51	0	**51**
PMP22	VIC	diagnostic	115	0	**115**	0	0	**0**
PMP22	WA	diagnostic	30	0	**30**	30	0	**30**
PMS2	SA	diagnostic	4	16	**20**	7	50	**57**
PMS2	SA	family	3	3	**6**	4	26	**30**
PMS2	VIC	family	4	0	**4**	4	1	**5**
PMS2	WA	diagnostic	2	0	**2**	19	0	**19**
PMS2	WA	family	0	0	**0**	2	0	**2**
POLG	VIC	diagnostic	0	0	**0**	17	7	**24**
POLG	VIC	family	3	3	**6**	2	15	**17**
POLG	WA	diagnostic	0	0	**0**	0	0	**0**
POU1F1	QLD	diagnostic	0	0	**0**	2	1	**3**
PPOX	NSW	diagnostic	0	0	**0**	4	1	**5**
PPP2R2B	WA	diagnostic	0	0	**0**	0	0	**0**
PPT1	SA	family	0	3	**3**	0	0	**0**
PRF1	WA	diagnostic	0	0	**0**	0	0	**0**
PRNP	VIC	diagnostic	21	31	**52**	10	28	**38**
PRNP	VIC	family	0	0	**0**	1	3	**4**
PRNP	VIC	screening	1	0	**1**	1	2	**3**
PRNP	WA	diagnostic	2	0	**2**	3	0	**3**

PROP1	QLD	diagnostic	0	0	**0**	2	1	**3**
PRPSAP2	SA	diagnostic	2	0	**2**	29	0	**29**
PRSS1	WA	diagnostic	4	0	**4**	3	0	**3**
PSEN1	VIC	diagnostic	30	10	**40**	10	11	**21**
PSEN1	VIC	family	0	0	**0**	0	2	**2**
PSEN2	VIC	diagnostic	0	0	**0**	2	1	**3**
PTEN	NSW	diagnostic	12	2	**14**	14	2	**16**
PTEN	NSW	family	0	0	**0**	4	1	**5**
PTEN	SA	diagnostic	1	0	**1**	1	0	**1**
PTEN	SA	family	4	0	**4**	0	0	**0**
PTEN	WA	diagnostic	4	0	**4**	5	0	**5**
PTPN11	WA	diagnostic	17	0	**17**	14	0	**14**
PYGM	WA	diagnostic	1	1	**2**	1	2	**3**
RAF1	WA	diagnostic	0	0	**0**	1	0	**1**
RAG1	WA	diagnostic	0	0	**0**	0	0	**0**
RAG2	WA	diagnostic	0	0	**0**	0	0	**0**
RAI1	SA	diagnostic	25	0	**25**	14	0	**14**
RAI1	WA	diagnostic	8	0	**8**	12	1	**13**
RARA	SA	somatic	9	0	**9**	13	1	**14**
RB1	NSW	somatic	0	0	**0**	60	1	**61**
RB1	SA	diagnostic	0	11	**11**	4	16	**20**
RB1	SA	family	0	13	**13**	6	12	**18**
RB1	SA	somatic	89	0	**89**	74	1	**75**
RB1	VIC	diagnostic	11	2	**13**	14	0	**14**
RB1	VIC	family	39	5	**44**	10	1	**11**
RB1	WA	diagnostic	8	0	**8**	7	0	**7**
RB1	WA	family	0	0	**0**	6	0	**6**
RET	NSW	diagnostic	21	5	**26**	13	6	**19**
RET	NSW	family	15	2	**17**	8	2	**10**
RET	SA	diagnostic	1	0	**1**	5	0	**5**
RET	VIC	diagnostic	65	10	**75**	103	14	**117**
RET	VIC	family	12	0	**12**	30	4	**34**
RET	WA	diagnostic	5	0	**5**	9	0	**9**
RET	WA	family	0	0	**0**	5	0	**5**
RHCE	SA	diagnostic	8	0	**8**	5	0	**5**

RHCE	VIC	diagnostic	10	0	**10**	0	0	**0**
RHCE	VIC	family	25	1	**26**	17	0	**17**
RHD	NSW	family	20	6	**26**	24	6	**30**
RHD	SA	diagnostic	8	0	**8**	5	0	**5**
RHD	VIC	diagnostic	10	0	**10**	0	0	**0**
RHD	VIC	family	25	1	**26**	17	0	**17**
RUNX1/RUNX1T1	SA	somatic	7	0	**7**	14	1	**15**
RUNX1/RUNX1T1	VIC	diagnostic	10	0	**10**	1	1	**2**
RUNX1/RUNX1T1	VIC	somatic	2	0	**2**	3	1	**4**
RUNX1/RUNX1T1	WA	somatic	11	0	**11**	11	0	**11**
RYR1	WA	diagnostic	4	13	**17**	5	7	**12**
SAMD12	SA	diagnostic	0	0	**0**	6	0	**6**
SBDS	WA	diagnostic	2	0	**2**	0	0	**0**
SCN1A	VIC	diagnostic	3	3	**6**	4	19	**23**
SCN1A	VIC	family	3	0	**3**	1	2	**3**
SCN4A	QLD	diagnostic	6	0	**6**	2	0	**2**
SCN4A	WA	diagnostic	3	1	**4**	45	30	**75**
SCN5A	VIC	diagnostic	34	5	**39**	35	13	**48**
SCN5A	VIC	family	9	0	**9**	14	1	**15**
SCN5A	WA	diagnostic	0	0	**0**	3	0	**3**
SCN5A	WA	family	0	0	**0**	1	0	**1**
SCNN1D	SA	diagnostic	2	0	**2**	29	0	**29**
SDHB	NSW	diagnostic	19	32	**51**	20	21	**41**
SDHB	NSW	family	18	27	**45**	2	18	**20**
SDHB	WA	diagnostic	0	0	**0**	6	0	**6**
SDHD	NSW	diagnostic	19	30	**49**	19	18	**37**
SDHD	NSW	family	0	7	**7**	4	7	**11**
SDHD	WA	diagnostic	0	0	**0**	6	0	**6**
SEMA5A	SA	diagnostic	0	0	**0**	3	0	**3**
sent o'seas#	ACT	diagnostic	22	0	**22**	0	0	**0**
sent o'seas#	NSW	diagnostic	5	0	**5**	10	0	**10**
sent o'seas#	QLD	supplementary	42	0	**42**	42	0	**42**
sent o'seas#	QLD	unknown	40	0	**40**	0	0	**0**
sent o'seas#	SA	diagnostic	60	0	**60**	17	35	**52**
sent o'seas#	SA	family	3	0	**3**	4	0	**4**

sent o'seas#	VIC	diagnostic	218	5	**223**	0	3	**3**
sent o'seas#	VIC	unknown	31	0	**31**	16	0	**16**
SEPN1	VIC	diagnostic	1	0	**1**	0	0	**0**
SEPN1	VIC	family	1	0	**1**	0	0	**0**
SERPINA1	QLD	diagnostic	482	0	**482**	482	0	**482**
SERPINA1	QLD	supplementary	5	0	**5**	5	0	**5**
SERPINA1	VIC	diagnostic	1	0	**1**	1	0	**1**
SERPINA1	VIC	family	5	0	**5**	1	0	**1**
SERPINA1	WA	diagnostic	795	0	**795**	867	1	**868**
SERPING1	NSW	diagnostic	0	30	**30**	0	0	**0**
SFTPB	NSW	diagnostic	2	0	**2**	0	21	**21**
SFTPB	NSW	family	2	0	**2**	0	22	**22**
SGSH	SA	diagnostic	0	2	**2**	0	1	**1**
SGSH	SA	family	0	6	**6**	0	4	**4**
SH2D1A	WA	diagnostic	2	0	**2**	3	0	**3**
SHOX	SA	diagnostic	18	0	**18**	2	0	**2**
SHOX	WA	diagnostic	0	0	**0**	4	0	**4**
SKI	SA	diagnostic	2	0	**2**	29	0	**29**
SLBP	SA	diagnostic	0	0	**0**	3	0	**3**
SLC14A1	SA	diagnostic	8	0	**8**	5	0	**5**
SLC14A1	VIC	diagnostic	10	0	**10**	0	0	**0**
SLC14A1	VIC	family	25	1	**26**	17	0	**17**
SLC14A1	WA	diagnostic	5	0	**5**	5	0	**5**
SLC26A4	WA	diagnostic	41	0	**41**	24	0	**24**
SLC26A4	WA	family	0	0	**0**	2	0	**2**
SMAD4	NSW	diagnostic	1	1	**2**	1	1	**2**
SMAD4	NSW	family	0	0	**0**	1	1	**2**
SMARCB1	VIC	somatic	2	0	**2**	12	1	**13**
SMCR	NSW	diagnostic	1	0	**1**	1	0	**1**
SMN1	NSW	diagnostic	66	20	**86**	56	20	**76**
SMN1	SA	diagnostic	16	2	**18**	17	2	**19**
SMN1	SA	family	0	0	**0**	3	1	**4**
SMN1	VIC	diagnostic	130	29	**159**	72	39	**111**
SMN1	VIC	family	321	87	**408**	59	76	**135**
SMN1	WA	diagnostic	26	0	**26**	15	0	**15**

SMPD1	VIC	screening	79	0	**79**	104	1	**105**
SNAP29	SA	diagnostic	2	0	**2**	29	0	**29**
SNCA	NSW	diagnostic	2	2	**4**	1	0	**1**
SNCA	VIC	diagnostic	0	3	**3**	0	5	**5**
SNCAIP	VIC	diagnostic	0	0	**0**	0	1	**1**
SNRPN	NSW	diagnostic	76	3	**79**	18	25	**43**
SNRPN	QLD	diagnostic	65	0	**65**	63	0	**63**
SNRPN	SA	diagnostic	46	5	**51**	62	4	**66**
SNRPN	VIC	diagnostic	178	0	**178**	0	0	**0**
SNRPN	WA	diagnostic	95	0	**95**	73	1	**74**
SOD1	NSW	diagnostic	11	7	**18**	16	0	**16**
SOD1	NSW	family	3	2	**5**	5	0	**5**
SOD1	VIC	diagnostic	1	0	**1**	5	1	**6**
SOD1	WA	diagnostic	4	6	**10**	10	12	**22**
Somatic hypermutation#	SA	somatic	1	0	**1**	2	4	**6**
SOS1	WA	diagnostic	0	0	**0**	11	0	**11**
SPAST	NSW	diagnostic	1	1	**2**	24	10	**34**
SPAST	NSW	family	0	0	**0**	0	0	**0**
SPAST	WA	diagnostic	11	0	**11**	12	0	**12**
SPG3A	NSW	diagnostic	1	1	**2**	24	10	**34**
SPG3A	NSW	family	0	0	**0**	0	0	**0**
SPG3A	WA	diagnostic	0	0	**0**	21	0	**21**
SPON2	SA	diagnostic	0	0	**0**	3	0	**3**
SPTLC1	NSW	diagnostic	13	5	**18**	13	2	**15**
SPTLC1	NSW	family	0	1	**1**	0	2	**2**
SRD5A2	WA	diagnostic	0	0	**0**	0	0	**0**
SRY	NSW	screening	20	6	**26**	24	6	**30**
SRY	QLD	diagnostic	334	0	**334**	334	0	**334**
SRY	SA	diagnostic	2	0	**2**	6	0	**6**
SRY	WA	diagnostic	4	0	**4**	3	1	**4**
SS18	SA	somatic	2	0	**2**	5	1	**6**
STK11	NSW	diagnostic	16	6	**22**	20	8	**28**
STK11	NSW	family	13	7	**20**	3	4	**7**
STK11	WA	diagnostic	0	0	**0**	1	0	**1**
STK11	WA	family	0	0	**0**	1	0	**1**

STR#	NSW	diagnostic	8	2	**10**	0	28	**28**
STR#	NSW	family	20	1	**21**	0	29	**29**
STR#	QLD	family	21	0	**21**	10	0	**10**
STR#	SA	family	19	1	**20**	15	1	**16**
STR#	SA	somatic	28	30	**58**	28	30	**58**
STR#	VIC	diagnostic	13	31	**44**	14	26	**40**
STR#	VIC	somatic	8	0	**8**	4	2	**6**
STS	SA	diagnostic	5	0	**5**	1	0	**1**
STX1A	SA	diagnostic	2	0	**2**	29	0	**29**
Subtel deletion#	NSW	diagnostic	131	0	**131**	489	2	**491**
Subtel deletion#	SA	diagnostic	749	0	**749**	837	0	**837**
Subtel deletion#	VIC	diagnostic	504	0	**504**	0	0	**0**
Subtel deletion#	WA	diagnostic	66	0	**66**	185	1	**186**
SURF1	VIC	diagnostic	2	2	**4**	3	0	**3**
SURF1	VIC	family	4	0	**4**	0	0	**0**
TACC3	SA	diagnostic	0	0	**0**	3	0	**3**
TBP	NSW	diagnostic	5	0	**5**	2	1	**3**
TBP	WA	diagnostic	0	0	**0**	0	0	**0**
TCF3	SA	somatic	8	9	**17**	10	9	**19**
TCF3/PBX1	QLD	somatic	120	0	**120**	94	0	**94**
TERT	SA	diagnostic	0	0	**0**	3	0	**3**
TGFBR1	NSW	diagnostic	2	3	**5**	0	27	**27**
TGFBR1	WA	diagnostic	0	0	**0**	0	0	**0**
TGFBR2	NSW	diagnostic	2	3	**5**	0	27	**27**
TGFBR2	WA	diagnostic	0	0	**0**	0	0	**0**
TH	VIC	diagnostic	0	0	**0**	0	1	**1**
TNFRSF13B	SA	diagnostic	2	0	**2**	29	0	**29**
TNFRSF18	SA	diagnostic	2	0	**2**	29	0	**29**
TNFRSF1A	NSW	diagnostic	58	44	**102**	0	25	**25**
TNFRSF1A	WA	family	0	0	**0**	1	0	**1**
TNFRSF4	SA	diagnostic	2	0	**2**	29	0	**29**
TOR1A	SA	diagnostic	5	6	**11**	4	8	**12**
TOR1A	SA	family	3	2	**5**	1	11	**12**
TOR1A	WA	diagnostic	6	12	**18**	7	14	**21**
TP53	NSW	somatic	7	0	**7**	42	1	**43**

TP53	SA	diagnostic	1	0	**1**	8	1	**9**
TP53	SA	family	0	0	**0**	1	1	**2**
TP53	SA	somatic	35	0	**35**	37	1	**38**
TP53	VIC	diagnostic	15	10	**25**	38	25	**63**
TP53	VIC	family	4	2	**6**	6	5	**11**
TP53	WA	diagnostic	1	0	**1**	14	0	**14**
TP53	WA	family	0	0	**0**	1	0	**1**
TP73	SA	diagnostic	2	0	**2**	29	0	**29**
TPM3	WA	diagnostic	1	2	**3**	0	0	**0**
TPMT	NSW	family	0	0	**0**	0	0	**0**
TPMT	SA	screening	41	1	**42**	64	1	**65**
TPMT	WA	screening	79	0	**79**	79	1	**80**
TPP1	SA	diagnostic	0	2	**2**	0	2	**2**
TPP1	SA	family	6	7	**13**	3	28	**31**
TPP1	SA	screening	2	2	**4**	2	7	**9**
TRAPPC2	SA	diagnostic	0	2	**2**	1	2	**3**
TRB@	QLD	somatic	955	0	**955**	1260	0	**1260**
TRB@	VIC	somatic	209	0	**209**	205	1	**206**
TRG@	QLD	somatic	955	0	**955**	1260	0	**1260**
TRG@	SA	diagnostic	101	3	**104**	162	3	**165**
TRG@	VIC	somatic	276	0	**276**	205	1	**206**
TRG@	WA	somatic	116	0	**116**	116	0	**116**
TRPS1	SA	diagnostic	5	0	**5**	8	0	**8**
TRPV1	SA	diagnostic	2	0	**2**	29	0	**29**
TSC1	NSW	diagnostic	1	3	**4**	30	3	**33**
TSC1	WA	diagnostic	9	0	**9**	12	0	**12**
TSC1	WA	family	0	0	**0**	0	0	**0**
TSC2	NSW	diagnostic	1	3	**4**	30	3	**33**
TSC2	VIC	family	4	0	**4**	0	0	**0**
TSC2	WA	diagnostic	9	0	**9**	12	0	**12**
TSC2	WA	family	0	0	**0**	0	0	**0**
TTN	VIC	diagnostic	0	0	**0**	2	0	**2**
TTR	NSW	diagnostic	2	0	**2**	2	1	**3**
TTR	VIC	diagnostic	1	0	**1**	0	1	**1**
TTR	VIC	family	2	0	**2**	8	1	**9**

TWIST1	NSW	diagnostic	50	56	**106**	46	41	**86**
TWIST1	NSW	family	5	0	**5**	0	0	**0**
TWIST1	SA	diagnostic	2	0	**2**	29	0	**29**
TWIST1	WA	diagnostic	0	0	**0**	22	0	**22**
TWIST1	WA	family	0	0	**0**	1	0	**1**
TWISTNB	SA	diagnostic	2	0	**2**	29	0	**29**
TYR	WA	diagnostic	1	0	**1**	1	0	**1**
TYR	WA	family	0	0	**0**	2	0	**2**
UBE3A	NSW	diagnostic	55	5	**60**	0	23	**23**
UBE3A	QLD	diagnostic	13	0	**13**	23	0	**23**
UBE3A	SA	diagnostic	45	0	**45**	68	1	**69**
UBE3A	VIC	diagnostic	178	0	**178**	0	0	**0**
UBE3A	WA	diagnostic	69	0	**69**	55	1	**56**
UCHL1	VIC	diagnostic	0	0	**0**	0	1	**1**
UGT1A1	NSW	family	0	0	**0**	0	0	**0**
UGT1A1	VIC	diagnostic	25	0	**25**	25	1	**26**
UGT1A1	VIC	screening	55	0	**55**	0	0	**0**
UGT1A1	WA	unknown	91	3	**94**	108	1	**109**
UNC13D	WA	family	3	0	**3**	2	0	**2**
VHL	NSW	diagnostic	15	19	**34**	12	10	**22**
VHL	NSW	family	1	0	**1**	1	2	**3**
VHL	SA	diagnostic	6	29	**35**	5	50	**54**
VHL	SA	family	4	15	**19**	1	25	**26**
VHL	WA	diagnostic	9	0	**9**	5	0	**5**
VHL	WA	family	0	0	**0**	0	0	**0**
VKORC1	VIC	pharmacogenetic	60	35	**95**	120	70	**190**
VWF	SA	diagnostic	5	0	**5**	5	0	**5**
WAS	WA	diagnostic	3	0	**3**	2	0	**2**
WAS	WA	family	0	0	**0**	0	0	**0**
WFS1	NSW	diagnostic	3	2	**5**	5	2	**7**
WFS1	NSW	family	1	0	**1**	2	1	**3**
WHSC1	SA	diagnostic	3	0	**3**	9	0	**9**
WHSC2	SA	diagnostic	0	0	**0**	3	0	**3**
WT1	SA	diagnostic	0	0	**0**	3	0	**3**
WT1	VIC	diagnostic	2	11	**13**	5	2	**7**

YWHAE	SA	diagnostic	3	0	**3**	0	0	**0**
YWHAE	WA	diagnostic	3	0	**3**	6	1	**7**

15.2.2 NUMBER OF LABORATORIES PROVIDING ACCREDITED AND NON-ACCREDITED TESTING, BY TYPE OF TEST AND BY REGION.

Type of test — HGNC test name, or non-standard name (indicated by #).
No. of labs offering accredited test — number of laboratories in the region reporting that the type of test was offered as an accredited test during 2006.
No. of labs offering non-accredited test — number of laboratories in the region reporting that the type of test was offered as a non-accredited test during 2006.
Accred unknown — number of laboratories in the region reporting that the type of test was offered during 2006 but without specifying accreditation status.

Tests which are offered as only non-accredited tests, or as both accredited and non-accredited tests, are highlighted by indent and **bold**.

	No. of labs offering accredited test							No. of labs offering non-accredited test							Accred unknown			GRAND TOTAL
Type of test	ACT	NSW	QLD	SA	VIC	WA	**TOTAL**	ACT	NSW	QLD	SA	VIC	WA	**TOTAL**	NSW	SA	**TOTAL**	
ABCC8			1				**1**											**1**
ABCD1				1			**1**											**1**
ABL1						1	**1**											**1**
ACADM		1	1	2	1	1	**6**											**6**
ACADS			1				**1**											**1**
ACADVL						1	**1**											**1**
ACTA1						1	**1**						1	**1**				**2**
ADA						1	**1**											**1**
ADSL			1				**1**											**1**
AFF2				1			**1**											**1**
ALDOB		1	1			1	**3**											**3**
ALK				1		1	**2**											**2**
ALPL						1	**1**											**1**
APC		1	1	2	1	1	**6**											**6**
APOB				1		1	**2**		1					**1**				**3**
APOE		1	2	1	1	1	**6**			1		1		**2**				**8**
APP					2		**2**											**2**
AR		1	2	1	1	2	**7**											**7**
ARSA				2			**2**											**2**
ARSE						1	**1**											**1**
ARVCF				1			**1**											**1**

ARX				1		1	**2**											**2**
ASPA		1		1	1		**3**											**3**
ATM		1		1		1	**3**											**3**
ATN1		1	1	1	1	1	**5**											**5**
ATP13A2					1		**1**											**1**
ATP1A2			1				**1**											**1**
ATP7A					1		**1**											**1**
ATRX						1	**1**											**1**
ATXN1		1	1	1	1	1	**5**											**5**
ATXN2		1	1	1	2	1	**6**											**6**
ATXN3		1	1	1	2	1	**6**											**6**
ATXN7		1	1	1	1	1	**5**											**5**
AVP			1				**1**											**1**
AZF#		1				1	**2**											**2**
BCHE			1			1	**2**											**2**
BCL2	1	1	2	1	1		**6**					1		**1**				**7**
BCL6						1	**1**											**1**
BCR/ABL1		1	1	3	2	1	**8**											**8**
BDNF				1			**1**											**1**
BLM		1			1		**2**											**2**
BMPR1A		1					**1**											**1**
BRAF		1		1	1	1	**4**											**4**
BRCA1		3	1	2	3	1	**10**											**10**
BRCA2		3	1	2	3	1	**10**											**10**
BTK						1	**1**											**1**
CACNA1A		1	2	1	1	1	**6**			1				**1**				**7**
CACNA1S			1			1	**2**											**2**
CASR			1				**1**						1	**1**				**2**
CAV3						1	**1**											**1**
CBFB		1		1			**2**											**2**
CBFB/MYH11						1	**1**					1		**1**				**2**
CCND1			1		1	1	**3**					1		**1**				**4**
CCND1/IGHG1		1		1		1	**3**											**3**
CCR5						1	**1**											**1**
CD109					1	1	**2**											**2**

CD177					1		**1**											**1**
CD2						1	**1**											**1**
CD40LG						1	**1**											**1**
CDC2L1				1		1	**2**											**2**
CDC45L																1	**1**	**1**
CDC73						1	**1**											**1**
CDH1		1			1		**2**		1					**1**				**3**
CDKN1B				1			**1**											**1**
CDKN1C					1		**1**											**1**
CDKN2A		1					**1**											**1**
CFTR	1	4	1	1	1	1	**9**					1		**1**				**10**
CHD7						1	**1**											**1**
Chimerism#		1		1	1	1	**4**											**4**
CLCN1						1	**1**											**1**
CLDN5																1	**1**	**1**
CLIP2																1	**1**	**1**
CLN3				1			**1**											**1**
CLTCL1																1	**1**	**1**
COL2A1					1		**1**											**1**
COL3A1		1					**1**											**1**
COL7A1			1				**1**											**1**
CPOX									1					**1**				**1**
CPS1					1	1	**2**											**2**
CREBBP				1		1	**2**											**2**
CRK				1		1	**2**											**2**
CTBP1				1			**1**											**1**
CTLA4					1		**1**											**1**
CTNND2				1			**1**											**1**
CTNS				1			**1**											**1**
CTSB				1			**1**											**1**
CYBB						1	**1**											**1**
CYP21A2			1			1	**2**											**2**
CYP2C19					1		**1**											**1**
CYP2C9				1	1		**2**											**2**
CYP2D6					1		**1**											**1**

D13S319#		1					**1**											**1**
D19S545#						1	**1**											**1**
D19S851#						1	**1**											**1**
D4Z4#									1				1	**2**				**2**
D5S721#				1			**1**											**1**
D7S613#				1		1	**2**											**2**
DARC				1	1	1	**3**											**3**
DCX						1	**1**											**1**
DDIT3		1		1			**2**											**2**
DGCR				1			**1**											**1**
DGCR2						1	**1**											**1**
DHCR7				1			**1**											**1**
DMD		1	1	1	1	1	**5**											**5**
DMPK		1	1	1	1	1	**5**											**5**
DYRK1A				1			**1**											**1**
EBP											1			**1**				**1**
EDA						1	**1**											**1**
EGFR				2	1	1	**4**											**4**
ELN		1		1		1	**3**									1	**1**	**4**
EMD					1		**1**											**1**
ERBB2				1			**1**											**1**
ETV6/RUNX1			1	1	1	1	**4**											**4**
EWSR1		1		1			**2**											**2**
EWSR1/FLI1					1		**1**											**1**
EWSR1/WT1					1		**1**											**1**
EXT1				1			**1**											**1**
F11				1			**1**											**1**
F12		1					**1**											**1**
F2			1				**1**											**1**
F5			1				**1**											**1**
F8			1	1	1	1	**4**		1					**1**				**5**
F9				1			**1**		1					**1**				**2**
FAH			1				**1**											**1**
FANCC		1			1		**2**											**2**
FBN1		1				1	**2**											**2**

FCGR3A					1		**1**											**1**
FDFT1				1			**1**											**1**
FGFR1		1			1	1	**3**											**3**
FGFR2		1			1	1	**3**											**3**
FGFR3		2	1	2	1	1	**7**											**7**
FGFRL1				1			**1**											**1**
FIP1L1		1					**1**											**1**
FIP1L1/PDGFRA		1					**1**					1		**1**				**2**
FKRP						1	**1**											**1**
FLCN		1				1	**2**											**2**
FLNA					1	1	**2**											**2**
FLT3						1	**1**					1		**1**				**2**
FMR1		1		1			**2**											**2**
FOXC2						1	**1**											**1**
FOXL2						1	**1**											**1**
FRAXA						1	**1**											**1**
FRAXE						1	**1**											**1**
FXN			1	1	1	1	**4**		1					**1**				**5**
FZD9				1			**1**											**1**
G6PC		1					**1**											**1**
GABRB3																1	**1**	**1**
GAK				1			**1**											**1**
GALT			1	1		1	**3**											**3**
GBA				1			**1**											**1**
GCH1					1		**1**											**1**
GCK			1				**1**											**1**
GDAP1		1					**1**											**1**
GFAP					1	1	**2**											**2**
GJB1		1				1	**2**											**2**
GJB2		2	1	1	1	1	**6**											**6**
GJB6		1		1	1	1	**4**											**4**
GLA				1			**1**											**1**
GLDC						1	**1**											**1**
GLUD1			1				**1**											**1**
GNB1																1	**1**	**1**

GP1BA					1	1	**2**											**2**
GRN					1		**1**											**1**
H19					1		**1**											**1**
HADHA			1	1		1	**3**											**3**
HBA1		2		1	1	2	**6**											**6**
HBA2		2		1	1	2	**6**											**6**
HBB		2		1	1	2	**6**		1					**1**				**7**
HBD					1		**1**		1					**1**				**2**
HBG1									1					**1**				**1**
HBG2									1					**1**				**1**
HEXA		1			1		**2**											**2**
HEXB		1					**1**											**1**
HFE		1	1		1		**3**											**3**
HIC1																1	**1**	**1**
HIPK3				1			**1**											**1**
HIRA		2	1	1		2	**6**											**6**
HLA-A					1	1	**2**											**2**
HLA-B				1	1	1	**3**		1					**1**				**4**
HLA-C					1	1	**2**											**2**
HLA-DPB1					1	1	**2**											**2**
HLA-DQA1					1		**1**								1		**1**	**2**
HLA-DQB1					1	1	**2**											**2**
HLA-DRB1				1	1	1	**3**								1		**1**	**4**
HLA-DRB3					1	1	**2**											**2**
HLA-DRB4						1	**1**											**1**
HLA-DRB5						1	**1**											**1**
HMBS									1					**1**				**1**
HMGCL						1	**1**											**1**
HMHA1					1		**1**											**1**
HNF1A			1				**1**											**1**
HNF4A										1				**1**				**1**
HPRT1			1				**1**											**1**
HRAS						1	**1**											**1**
HSD17B4				1			**1**											**1**
HTT		1	1	1	1	1	**5**											**5**

IDS				1			**1**											**1**
IDUA				1			**1**											**1**
IGH@	1		2	2	1	1	**7**											**7**
IGH@/BCL2				1		2	**3**											**3**
IGH@/CCND1				1		1	**2**											**2**
IGH@/MYC						1	**1**											**1**
IGHG1		1				1	**2**											**2**
IGHR												1		**1**				**1**
IKBKAP		1			1		**2**											**2**
IKBKG				1		1	**2**											**2**
IL2RG		1				1	**2**											**2**
IL7R						1	**1**											**1**
ITGA2					1	1	**2**											**2**
ITGA2B					1	1	**2**											**2**
ITGB3					1	1	**2**											**2**
ITGB4			1				**1**											**1**
JAG1		1			1	1	**3**									1	**1**	**4**
JAK2		1		2	1	1	**5**		1	1		1		**3**				**8**
KAL1				1		1	**2**											**2**
KCNE1					1	1	**2**											**2**
KCNE2					1	1	**2**											**2**
KCNH2					1	1	**2**											**2**
KCNJ11			1				**1**											**1**
KCNJ2					1		**1**											**1**
KCNJ6				1			**1**											**1**
KCNQ1					1	1	**2**											**2**
KCNQ1OT1					1		**1**											**1**
KEL				1	1	1	**3**											**3**
KIR3DL1					1		**1**											**1**
KIT		1			1		**2**											**2**
KMS				1			**1**											**1**
KRAS					1	1	**2**											**2**
KRIT1						1	**1**											**1**
KRT10					1		**1**											**1**
KRT14			1				**1**											**1**

KRT5			1				**1**											**1**
L1CAM					1	1	**2**											**2**
LAMA3			1				**1**											**1**
LAMB3			1				**1**											**1**
LAMC2			1				**1**											**1**
LDLR									1					**1**				**1**
LETM1				1			**1**											**1**
LIMK1				1		1	**2**									1	**1**	**3**
LLGL1																1	**1**	**1**
LMNA		1			1		**2**											**2**
LRRK2					1		**1**											**1**
MAPT					1	1	**2**											**2**
MAT1A						1	**1**											**1**
MBP						1	**1**											**1**
MC2R			1				**1**											**1**
MC4R			1				**1**											**1**
MCOLN1		1					**1**											**1**
MECP2		1				1	**2**			1				**1**				**3**
MEFV		2		1		1	**4**											**4**
MEN1			1	1	1	1	**4**											**4**
MET		1		1			**2**											**2**
MFAP4																1	**1**	**1**
MFN2									1				1	**2**				**2**
MGMT												1		**1**				**1**
MKRN3																1	**1**	**1**
MLH1		1	1	2	3	1	**8**											**8**
MLL		1		1			**2**											**2**
MLL/AFF1			1				**1**											**1**
MNX1						1	**1**											**1**
MPZ		1				1	**2**											**2**
MSH2		1	1	2	3	1	**8**											**8**
MSH6		1		2	3	1	**7**											**7**
MSI#				1	3	1	**5**		1					**1**				**6**
MSRA				1			**1**											**1**
MT-ATP6			1	1	2		**4**											**4**

MT-CO1					1		**1**											**1**
MT-CO2					1		**1**											**1**
MT-CO3					1		**1**											**1**
MT-CYB			1				**1**											**1**
MT-deletion#			1		1		**2**											**2**
MTHFR			4		1		**5**											**5**
MTM1						1	**1**											**1**
MT-ND1			1	1	1		**3**											**3**
MT-ND4			1	1	1		**3**											**3**
MT-ND5		1		1	1		**3**											**3**
MT-ND6				1	1		**2**											**2**
MT-RNR1				1	1	1	**3**											**3**
MT-TK		1	1	1	2		**5**											**5**
MT-TL1			1	1	1		**3**											**3**
MUTYH		1		2	1	1	**5**											**5**
MYBPC3					1		**1**											**1**
MYC		1		1		1	**3**											**3**
MYCN			1		1	1	**3**											**3**
MYH7					1		**1**						1	**1**				**2**
MYOT													1	**1**				**1**
NAGLU				1			**1**											**1**
NDN																1	**1**	**1**
NDP			1			1	**2**											**2**
NF1						1	**1**											**1**
NF2				1	1	1	**3**											**3**
NIPA1									1					**1**				**1**
NOD2								1				1		**2**				**2**
NOG						1	**1**											**1**
NOTCH3			1			1	**2**											**2**
NPC1		1		1			**2**											**2**
NR0B1						1	**1**											**1**
NR4A2					1		**1**											**1**
NSD1				1		1	**2**									1	**1**	**3**
OCRL						1	**1**											**1**
OTC		1	1	1	1		**4**											**4**

OTOF						1	**1**											**1**
PABPN1			1				**1**											**1**
PAFAH1B1		1		1		2	**4**									1	**1**	**5**
PAH			1				**1**											**1**
PALB2						1	**1**											**1**
PANK2					1		**1**											**1**
PARK2					1		**1**											**1**
PARK7					1		**1**											**1**
PAX6				1			**1**											**1**
PAX7/FOXO1A					1		**1**											**1**
PDCD6				1			**1**											**1**
PDHA1					1		**1**											**1**
PEX1				1			**1**											**1**
PEX7				1			**1**											**1**
PINK1					1		**1**											**1**
PITX2						1	**1**											**1**
PKP2					1		**1**											**1**
PLA2G6						1	**1**											**1**
PML/RARA		1		2	2	1	**6**											**6**
PMM2				1			**1**											**1**
PMP22		1		1	1	1	**4**											**4**
PMS2				1	1	1	**3**											**3**
POLG					1	1	**2**											**2**
POU1F1			1				**1**											**1**
PPOX									1					**1**				**1**
PPP2R2B						1	**1**											**1**
PPT1				1			**1**											**1**
PRF1						1	**1**											**1**
PRNP					2	1	**3**					1		**1**				**4**
PROP1			1				**1**											**1**
PRPSAP2																1	**1**	**1**
PRSS1						1	**1**											**1**
PSEN1					2		**2**											**2**
PSEN2					1		**1**											**1**
PTEN		1		1		1	**3**											**3**

PTPN11						1	**1**											**1**
PYGM						1	**1**											**1**
RAF1						1	**1**											**1**
RAG1						1	**1**											**1**
RAG2						1	**1**											**1**
RAI1				1		1	**2**											**2**
RARA				1			**1**											**1**
RB1		1		2	1	1	**5**											**5**
RET		1		1	2	1	**5**											**5**
RHCE				1	1		**2**											**2**
RHD				1	1		**2**		1					**1**				**3**
RUNX1/RUNX1T1				1	2	1	**4**											**4**
RYR1						1	**1**											**1**
SAMD12				1			**1**											**1**
SBDS						1	**1**											**1**
SCN1A					1		**1**											**1**
SCN4A			1			1	**2**											**2**
SCN5A					1	1	**2**											**2**
SCNN1D																1	**1**	**1**
SDHB		1				1	**2**											**2**
SDHD		1				1	**2**											**2**
SEMA5A				1			**1**											**1**
SEPN1					1		**1**											**1**
SERPINA1			1		1	2	**4**											**4**
SERPING1															1		**1**	**1**
SFTPB		1					**1**											**1**
SGSH				1			**1**											**1**
SH2D1A						1	**1**											**1**
SHOX				1		1	**2**											**2**
SKI																1	**1**	**1**
SLBP				1			**1**											**1**
SLC14A1				1	1	1	**3**											**3**
SLC26A4						1	**1**											**1**
SMAD4		1					**1**											**1**
SMARCB1					1		**1**											**1**

SMCR		1					**1**											**1**
SMN1		1		1	1	1	**4**											**4**
SMPD1					1		**1**											**1**
SNAP29																1	**1**	**1**
SNCA		1			1		**2**											**2**
SNCAIP					1		**1**											**1**
SNRPN		3	2	2	1	2	**10**											**10**
SOD1		1			1	1	**3**											**3**
Somatic hypermutation#				1			**1**											**1**
SOS1						1	**1**											**1**
SPAST						1	**1**		1					**1**				**2**
SPG3A									1				1	**2**				**2**
SPON2				1			**1**											**1**
SPTLC1		1					**1**											**1**
SRD5A2						1	**1**											**1**
SRY			1	1		1	**3**		1	1				**2**				**5**
SS18				1			**1**											**1**
STK11		1				1	**2**											**2**
STR#		1	1	2	2		**6**											**6**
STS				1			**1**											**1**
STX1A																1	**1**	**1**
Subtel deletion#		1		1	1	1	**4**		1					**1**				**5**
SURF1					1		**1**											**1**
TACC3				1			**1**											**1**
TBP						1	**1**		1					**1**				**2**
TCF3				1			**1**											**1**
TCF3/PBX1			1				**1**											**1**
TERT				1			**1**											**1**
TGFBR1		1				1	**2**											**2**
TGFBR2		1				1	**2**											**2**
TH					1		**1**											**1**
TNFRSF13B																1	**1**	**1**
TNFRSF18																1	**1**	**1**
TNFRSF1A		2				1	**3**											**3**
TNFRSF4																1	**1**	**1**

TOR1A				1		1	**2**										**2**
TP53		1		2	1	1	**5**										**5**
TP73															1	**1**	**1**
TPM3						1	**1**										**1**
TPMT				1		1	**2**		1					**1**			**3**
TPP1				1			**1**										**1**
TRAPPC2				1			**1**										**1**
TRB@			2		1		**3**										**3**
TRG@			2	1	1	1	**5**					1		**1**			**6**
TRPS1				1			**1**										**1**
TRPV1															1	**1**	**1**
TSC1						1	**1**		1					**1**			**2**
TSC2					1	1	**2**		1					**1**			**3**
TTN					1		**1**										**1**
TTR		1			2		**3**										**3**
TWIST1		1				1	**2**								1	**1**	**3**
TWISTNB															1	**1**	**1**
TYR						1	**1**										**1**
UBE3A		1	1	2	1	2	**7**								1	**1**	**8**
UCHL1					1		**1**										**1**
UGT1A1					1	1	**2**		1			1		**2**			**4**
UNC13D						1	**1**										**1**
VHL		1		1		1	**3**										**3**
VKORC1					1		**1**										**1**
VWF				1			**1**										**1**
WAS						1	**1**										**1**
WFS1		1					**1**										**1**
WHSC1				1			**1**										**1**
WHSC2				1			**1**										**1**
WT1				1	1		**2**										**2**
YWHAE				1		1	**2**										**2**

15.2.3 METHODS USED FOR DIAGNOSTIC TESTING BY TYPE OF TEST

This table lists by type of test the number of laboratories using the specified method for diagnostic testing.

Type of test HGNC test name, or non-standard name (indicated by #).
FISH fluorescent in situ hybridisation; other methods as listed

Type of test	FISH	mutation screen	segregation study	sequencing	sequencing plus MLPA	Southern	specific assay/s	TOTAL
ABCC8				1				1
ABCD1				1			1	2
ACADM				2			2	4
ACADS				1				1
ACTA1				2				2
ADA				1				1
ADSL				1				1
AFF2							1	1
ALDOB				2			1	3
ALPL				1				1
APC				1	5			6
APOB				1			2	3
APOE		1					4	5
APP		1		1				2
AR				2			5	7
ARSA	1			1				2
ARSE				1				1
ARVCF							1	1
ARX		1					1	2
ASPA							2	2
ATM					1			1
ATN1							5	5
ATP13A2		1						1
ATP1A2				1				1
ATRX		1						1
ATXN1							5	5
ATXN2		1					5	6
ATXN3		1					5	6
ATXN7							5	5

AVP				1				1
AZF#							2	2
BCHE				1				1
BDNF							1	1
BLM							1	1
BMPR1A				1				1
BRAF				1				1
BRCA1		1		3	8		3	15
BRCA2		1		3	8		3	15
BTK				1				1
CACNA1A				3			5	8
CACNA1S		1					1	2
CASR				2				2
CAV3				1				1
CD109							1	1
CD2							1	1
CD40LG				1				1
CDC2L1	2							2
CDC45L							1	1
CDC73				1				1
CDH1				2				2
CDKN1B	1							1
CDKN1C		1						1
CFTR					1		6	7
CHD7					1			1
Chimerism#							1	1
CLCN1				1				1
CLDN5							1	1
CLIP2							1	1
CLN3				1			1	2
CLTCL1							1	1
COL7A1				1				1
CPOX				1				1

CPS1				1				1
CREBBP	1						1	2
CRK	2							2
CTBP1							1	1
CTNND2							1	1
CTNS							1	1
CTSB							1	1
CYBB				2				2
CYP21A2					2			2
D4Z4#						2		2
D5S721#	1							1
D7S613#	2							2
DARC							3	3
DCX					1			1
DGCR							1	1
DGCR2	1							1
DHCR7				1			1	2
DMD			1				4	5
DMPK				1		1	3	5
DYRK1A							1	1
EBP				1				1
EDA				1				1
ELN	3						1	4
EMD				1				1
EXT1							1	1
F11				1				1
F12							1	1
F2							1	1
F5							1	1
F8				1	2	1	2	6
F9				2				2
FAH				1				1
FANCC							1	1

FBN1		1			1			2
FDFT1							1	1
FGFR1					2		1	3
FGFR2					2		1	3
FGFR3					2		5	7
FGFRL1							1	1
FKRP				1				1
FLNA				1				1
FMR1							1	1
FOXC2					1			1
FOXL2					1			1
FRAXA							1	1
FRAXE							1	1
FXN				1			4	5
FZD9							1	1
G6PC							1	1
GABRB3							1	1
GAK							1	1
GALT		1					2	3
GBA							1	1
GCH1		1						1
GCK				1				1
GDAP1				1				1
GFAP				1				1
GJB1				2				2
GJB2				4			3	7
GJB6				1			3	4
GLA				1			1	2
GLDC				1				1
GLUD1				1				1
GNB1							1	1
GP1BA							1	1
GRN		1						1

H19							1	1
HADHA				2			1	3
HBA1		1			1		4	6
HBA2		1			1		4	6
HBB		1		2	1		2	6
HBD							2	2
HBG1					1			1
HBG2					1			1
HEXA							2	2
HEXB							1	1
HFE							3	3
HIC1							1	1
HIPK3							1	1
HIRA	6							6
HLA-A				2				2
HLA-B				1				1
HLA-C				1				1
HLA-DQA1							1	1
HLA-DRB1				1			1	2
HLA-DRB3				1				1
HMBS				1				1
HMGCL				1				1
HNF1A					1			1
HNF4A					1			1
HPRT1				1				1
HRAS				1				1
HSD17B4							1	1
HTT			1				4	5
IDS				1				1
IGH@							1	1
IKBKAP							1	1
IKBKG		1					1	2
IL2RG				1			1	2

IL7R				1				1
ITGA2							1	1
ITGA2B							2	2
ITGB3							1	1
ITGB4				1				1
JAG1		1		1			1	3
JAK2							1	1
KAL1	1				1			2
KCNE1				1	1			2
KCNE2				1	1			2
KCNH2				1	1			2
KCNJ11				1				1
KCNJ2					1			1
KCNJ6							1	1
KCNQ1				1	1			2
KCNQ1OT1							1	1
KEL							3	3
KMS	1							1
KRAS				1				1
KRIT1					1			1
KRT14				1				1
KRT5				1				1
L1CAM				1				1
LAMA3				1				1
LAMB3				1				1
LAMC2				1				1
LDLR					1			1
LETM1							1	1
LIMK1	2						1	3
LLGL1							1	1
LMNA				4				4
LRRK2		1						1
MAPT		1		1				2

MAT1A		1						1
MBP		1						1
MC2R				1				1
MC4R				1				1
MCOLN1							1	1
MECP2					3			3
MEFV		1		1			1	3
MEN1				2	1		1	4
MET				1				1
MFAP4							1	1
MFN2		1		1				2
MKRN3							1	1
MLH1				1	7		1	9
MNX1				1				1
MPZ				2				2
MSH2				1	7		1	9
MSH6					7			7
MSI#							3	3
MSRA							1	1
MT-ATP6				1			3	4
MT-CO1				1				1
MT-CO2				1				1
MT-CO3				1				1
MT-CYB							1	1
MT-deletion#		1					1	2
MTHFR		1					2	3
MTM1				1				1
MT-ND1				1			2	3
MT-ND4				1			2	3
MT-ND5				1			2	3
MT-ND6				1			1	2
MT-RNR1				3				3
MT-TK				1			4	5

MT-TL1				1			2	3
MUTYH				2	2		1	5
MYBPC3				1				1
MYCN				1				1
MYH7				2				2
MYOT				1				1
NDN							1	1
NDP				1			1	2
NF1					1			1
NF2					3			3
NIPA1		1						1
NOD2				1				1
NOG				1				1
NOTCH3				2				2
NPC1							2	2
NR0B1				1				1
NR4A2		1						1
NSD1	1				1		1	3
OCRL				1				1
OTC				1	1			2
OTOF		1						1
PABPN1							1	1
PAFAH1B1	3				1		1	5
PAH							1	1
PALB2							1	1
PANK2		1						1
PARK2		1						1
PARK7		1						1
PAX6	1						1	2
PDCD6							1	1
PINK1		1						1
PITX2					1			1
PLA2G6				1				1

PMM2				1			1	2
PMP22					2		2	4
PMS2					2			2
POLG		1					1	2
POU1F1				1				1
PPOX				1				1
PPP2R2B							1	1
PRF1				1				1
PRNP		1		3			2	6
PROP1				1				1
PRPSAP2							1	1
PRSS1							1	1
PSEN1		1		1				2
PSEN2		1						1
PTEN				2	1			3
PTPN11				1				1
PYGM				1				1
RAF1				1				1
RAG1				1				1
RAG2				1				1
RAI1	2							2
RB1					5			5
RET				5	1			6
RHCE							2	2
RHD							2	2
RUNX1/RUNX1T1		1						1
RYR1				1				1
SAMD12							2	2
SBDS		1						1
SCN1A				1				1
SCN4A				1			1	2
SCN5A				1	1			2
SCNN1D							1	1

SDHB				1	1			2
SDHD				1	1			2
SEMA5A							1	1
SEPN1				1				1
SERPINA1				1			3	4
SERPING1				1				1
SFTPB							1	1
SGSH				1			1	2
SH2D1A				1				1
SHOX	1						1	2
SKI							1	1
SLBP							1	1
SLC14A1							3	3
SLC26A4					1			1
SMAD4				1				1
SMCR	1							1
SMN1					1		3	4
SNAP29							1	1
SNCA		1					1	2
SNCAIP		1						1
SNRPN	6					1	4	11
SOD1		1		2				3
SOS1				1				1
SPAST		1			1			2
SPG3A		1					1	2
SPON2							1	1
SPTLC1				1				1
SRD5A2				1				1
SRY	3						2	5
STK11					2			2
STR#							2	2
STS	1							1
STX1A							1	1

Subtel deletion#	3						3	6
SURF1				1				1
TACC3							1	1
TBP				1			1	2
TERT							1	1
TGFBR1		1			1			2
TGFBR2		1			1			2
TH		1						1
TNFRSF13B							1	1
TNFRSF18							1	1
TNFRSF1A		1		1				2
TNFRSF4							1	1
TOR1A							2	2
TP53				1	2			3
TP73							1	1
TPM3				1				1
TPP1				1			1	2
TRAPPC2				1				1
TRG@							1	1
TRPS1	1						2	3
TRPV1							1	1
TSC1					2			2
TSC2					2			2
TTN							1	1
TTR		1		1				2
TWIST1					2		1	3
TWISTNB							1	1
TYR				1				1
UBE3A	2					1	5	8
UCHL1		1						1
UGT1A1							1	1
VHL				1	2			3
VWF				1				1

WAS				1				1
WFS1				1				1
WHSC1	1						1	2
WHSC2							1	1
WT1		1					1	2
YWHAE	2							2

15.2.4 RATE OF TESTING BY TYPE OF TEST AND REGION

Presented as number of assays per million population. Tests performed on samples from outside the testing region ("interstate") were apportioned to other regions in proportion to their population.

Type of test HGNC test name, or non-standard name (indicated by #).
Region as listed

Type of test	ACT	NSW	NT	QLD	SA	TAS	VIC	WA	National
ABCC8	0.25	0.25	0.25	0.00	0.25	0.25	0.25	0.25	**0.20**
ABCD1	1.19	1.19	1.19	1.19	5.22	1.19	1.19	1.19	**1.49**
ABL1	0.00	0.00	0.00	0.00	0.00	0.00	0.00	4.04	**0.40**
ACADM	1.78	2.22	1.78	12.32	35.88	1.78	4.20	6.83	**7.67**
ACADS	0.00	0.00	0.00	0.00	0.00	0.00	0.00	0.00	**0.00**
ACADVL	0.00	0.00	0.00	0.00	0.00	0.00	0.00	0.00	**0.00**
ACTA1	1.55	1.55	1.55	1.55	1.55	1.55	1.55	1.01	**1.49**
ADA	0.00	0.00	0.00	0.00	0.00	0.00	0.00	0.00	**0.00**
ADSL	0.00	0.00	0.00	0.00	0.00	0.00	0.00	0.00	**0.00**
AFF2	0.22	0.22	0.22	0.22	3.91	0.22	0.22	0.22	**0.50**
ALDOB	0.45	0.30	0.45	0.71	0.45	0.45	0.45	1.46	**0.55**
ALK	0.00	0.00	0.00	0.00	3.26	0.00	0.00	7.58	**1.00**
ALPL	0.00	0.00	0.00	0.00	0.00	0.00	0.00	0.00	**0.00**
APC	4.16	11.90	4.16	16.25	30.32	4.16	28.20	10.73	**17.67**
APOB	0.00	0.00	0.00	0.00	3.91	0.00	0.00	10.11	**1.29**
APOE	0.00	8.93	0.00	97.73	234.88	0.00	93.09	50.55	**67.80**
APP	0.66	0.66	0.66	0.66	0.66	0.66	6.25	0.66	**2.04**
AR	1.11	4.68	1.11	8.73	14.16	1.11	6.96	9.20	**7.02**

ARSA	0.32	0.32	0.32	0.32	3.26	0.32	0.32	0.32	**0.55**
ARSE	0.00	0.00	0.00	0.00	0.00	0.00	0.00	0.00	**0.00**
ARVCF	0.00	0.00	0.00	0.00	1.30	0.00	0.00	0.00	**0.10**
ARX	0.11	0.11	0.11	0.11	12.40	0.11	0.11	0.61	**1.10**
ASPA	0.00	55.05	0.00	0.00	0.00	0.00	15.92	0.00	**22.35**
ATM	0.00	1.04	0.00	0.00	22.84	0.00	0.00	0.51	**2.14**
ATN1	0.12	0.42	0.12	32.53	30.79	0.12	1.47	1.58	**9.31**
ATP13A2	0.00	0.00	0.00	0.00	0.00	0.00	0.00	0.00	**0.00**
ATP1A2	0.00	0.00	0.00	0.00	0.00	0.00	0.00	0.00	**0.00**
ATP7A	0.00	0.00	0.00	0.00	0.00	0.00	2.42	0.00	**0.60**
ATRX	0.00	0.00	0.00	0.00	0.00	0.00	0.00	0.00	**0.00**
ATXN1	0.29	7.80	0.29	32.70	30.95	0.29	23.60	27.08	**19.81**
ATXN2	0.29	8.70	0.29	32.70	30.95	0.29	23.19	27.08	**20.01**
ATXN3	0.29	8.10	0.29	32.70	30.95	0.29	23.19	29.11	**20.01**
ATXN7	0.29	8.84	0.29	32.70	30.95	0.29	23.19	28.10	**20.16**
AVP	0.00	0.00	0.00	0.00	0.00	0.00	0.00	0.00	**0.00**
AZF#	0.00	13.84	0.00	0.00	0.00	0.00	0.00	4.55	**5.08**
BCHE	0.00	0.00	0.00	3.86	0.00	0.00	0.00	11.12	**1.84**
BCL2	49.36	1.93	0.00	245.62	2.61	0.00	26.80	0.00	**55.80**
BCL6	0.00	0.00	0.00	0.00	0.00	0.00	0.00	6.57	**0.65**
BCR/ABL1	61.81	63.30	61.81	92.68	639.38	61.81	162.76	67.88	**137.89**
BCR/ABL1 [MBS]	49.36	85.85	25.02	100.82	173.55	18.66	49.16	61.68	**81.19**
BDNF	0.00	0.00	0.00	0.00	0.00	0.00	0.00	0.00	**0.00**
BLM	0.00	8.48	0.00	0.00	0.00	0.00	15.92	0.00	**6.77**
BMPR1A	0.22	0.45	0.22	0.22	0.22	0.22	0.22	0.22	**0.30**
BRAF	0.13	15.01	0.13	0.13	0.78	0.13	1.41	0.13	**5.48**
BRCA1	12.39	70.56	12.39	38.37	106.07	12.39	53.20	65.98	**59.39**
BRCA2	13.52	69.76	13.52	39.49	99.32	13.52	49.98	64.58	**57.94**
BTK	0.00	0.00	0.00	0.00	0.00	0.00	0.00	1.01	**0.10**
CACNA1A	1.65	9.76	1.65	33.21	35.57	1.65	24.55	30.46	**21.55**
CACNA1S	0.11	0.11	0.11	1.65	0.11	0.11	0.11	3.03	**0.70**
CASR	0.11	0.11	0.11	2.43	0.11	0.11	0.11	5.56	**1.10**
CAV3	0.00	0.00	0.00	0.00	0.00	0.00	0.00	0.51	**0.05**

CBFB	0.00	0.89	0.00	0.00	3.91	0.00	0.00	0.00	**0.60**
CBFB/MYH11	0.00	0.00	0.00	0.00	0.00	0.00	1.41	6.57	**1.00**
CCND1	0.00	0.00	0.00	142.49	0.00	0.00	4.03	1.52	**28.72**
CCND1/IGHG1	0.00	4.02	0.00	0.00	51.54	0.00	0.00	2.53	**5.53**
CCR5	0.00	0.00	0.00	0.00	0.00	0.00	0.00	22.24	**2.19**
CD109	1.32	1.32	1.32	1.32	1.32	1.32	229.70	16.49	**59.24**
CD177	0.00	0.00	0.00	0.00	0.00	0.00	2.01	0.00	**0.50**
CD2	0.00	0.00	0.00	0.00	0.00	0.00	0.00	0.00	**0.00**
CD40LG	0.00	0.00	0.00	0.00	0.00	0.00	0.00	0.00	**0.00**
CDC2L1	0.00	0.00	0.00	0.00	11.09	0.00	0.00	3.03	**1.14**
CDC45L	0.00	0.00	0.00	0.00	1.30	0.00	0.00	0.00	**0.10**
CDC73	0.00	0.00	0.00	0.00	0.00	0.00	0.00	0.51	**0.05**
CDH1	0.37	0.89	0.37	0.37	0.37	0.37	0.37	0.37	**0.55**
CDKN1B	0.00	0.00	0.00	0.00	1.96	0.00	0.00	0.00	**0.15**
CDKN1C	0.00	0.00	0.00	0.00	0.00	0.00	0.00	0.00	**0.00**
CDKN2A	0.15	0.15	0.15	0.15	0.15	0.15	0.15	0.15	**0.15**
CFTR	116.60	739.54	20.95	607.36	416.40	20.95	496.98	307.09	**552.35**
CHD7	0.00	0.00	0.00	0.00	0.00	0.00	0.00	0.51	**0.05**
Chimerism#	0.00	0.15	0.00	0.00	7.83	0.00	0.00	16.68	**2.29**
CLCN1	0.83	0.83	0.83	0.83	0.83	0.83	0.83	3.03	**1.05**
CLDN5	0.00	0.00	0.00	0.00	1.30	0.00	0.00	0.00	**0.10**
CLIP2	0.00	0.00	0.00	0.00	1.30	0.00	0.00	0.00	**0.10**
CLN3	1.35	1.35	1.35	1.35	1.30	1.35	1.35	1.35	**1.34**
CLTCL1	0.00	0.00	0.00	0.00	1.30	0.00	0.00	0.00	**0.10**
COL2A1	0.00	0.00	0.00	0.00	0.00	0.00	0.60	0.00	**0.15**
COL3A1	0.00	0.60	0.00	0.00	0.00	0.00	0.00	0.00	**0.20**
COL7A1	0.00	0.00	0.00	2.31	0.00	0.00	0.00	0.00	**0.45**
CPOX	0.00	0.00	0.00	0.00	0.00	0.00	0.00	0.00	**0.00**
CPS1	0.00	0.00	0.00	0.00	0.00	0.00	0.00	0.00	**0.00**
CREBBP	0.39	0.39	0.39	0.39	0.39	0.39	0.39	0.51	**0.40**
CRK	0.00	0.00	0.00	0.00	1.96	0.00	0.00	1.52	**0.30**
CTBP1	0.00	0.00	0.00	0.00	0.00	0.00	0.00	0.00	**0.00**
CTLA4	0.00	0.00	0.00	0.00	0.00	0.00	4.03	0.00	**1.00**

CTNND2	0.00	0.00	0.00	0.00	0.00	0.00	0.00	0.00	**0.00**
CTNS	0.22	0.22	0.22	0.22	1.96	0.22	0.22	0.22	**0.35**
CTSB	0.00	0.00	0.00	0.00	0.00	0.00	0.00	0.00	**0.00**
CYBB	0.00	0.00	0.00	0.00	0.00	0.00	0.00	0.51	**0.05**
CYP21A2	1.79	1.79	1.79	2.83	1.79	1.79	1.79	7.86	**2.59**
CYP2C19	2.64	2.64	2.64	2.64	2.64	2.64	20.15	2.64	**6.97**
CYP2C9	2.31	2.31	2.31	2.31	2.31	2.31	12.09	2.31	**4.73**
CYP2D6	2.64	2.64	2.64	2.64	2.64	2.64	20.15	2.64	**6.97**
D13S319#	0.00	0.00	0.00	0.00	0.00	0.00	0.00	0.00	**0.00**
D19S545#	0.00	0.00	0.00	0.00	0.00	0.00	0.00	16.18	**1.59**
D19S851#	0.00	0.00	0.00	0.00	0.00	0.00	0.00	16.18	**1.59**
D4Z4#	3.65	8.69	3.65	3.65	3.65	3.65	3.65	19.77	**6.92**
D5S721#	0.00	0.00	0.00	0.00	0.65	0.00	0.00	0.00	**0.05**
D7S613#	0.00	0.00	0.00	0.00	9.79	0.00	0.00	7.58	**1.49**
DARC	0.07	0.07	0.07	0.07	5.29	0.07	7.05	2.59	**2.44**
DCX	0.61	0.61	0.61	0.61	0.61	0.61	0.61	1.01	**0.65**
DDIT3	0.00	0.00	0.00	0.00	1.96	0.00	0.00	0.00	**0.15**
DGCR	0.00	0.00	0.00	0.00	3.26	0.00	0.00	0.00	**0.25**
DGCR2	0.00	0.00	0.00	0.00	0.00	0.00	0.00	2.53	**0.25**
DHCR7	0.32	0.32	0.32	0.32	0.00	0.32	0.32	0.32	**0.30**
DMD	5.87	25.34	5.87	9.73	20.12	5.87	49.87	11.43	**25.64**
DMPK	3.03	14.79	3.03	23.86	44.74	3.03	65.93	15.67	**30.96**
DYRK1A	0.00	0.00	0.00	0.00	0.00	0.00	0.00	0.00	**0.00**
EBP	0.00	0.00	0.00	0.00	1.30	0.00	0.00	0.00	**0.10**
EDA	0.00	0.00	0.00	0.00	0.00	0.00	0.00	0.00	**0.00**
EGFR	3.41	3.41	3.41	3.41	9.94	3.41	28.70	19.59	**11.75**
ELN	0.00	0.15	0.00	0.00	11.09	0.00	0.00	7.58	**1.64**
EMD	0.79	0.79	0.79	0.79	0.79	0.79	5.64	0.79	**1.99**
ERBB2	0.00	0.00	0.00	0.00	0.00	0.00	0.00	0.00	**0.00**
ETV6/RUNX1	0.49	0.49	0.49	31.35	4.57	0.49	4.31	6.55	**8.31**
EWSR1	0.00	0.45	0.00	0.00	5.87	0.00	0.00	0.00	**0.60**
EWSR1/FLI1	0.00	0.00	0.00	0.00	0.00	0.00	0.60	0.00	**0.15**
EWSR1/WT1	0.00	0.00	0.00	0.00	0.00	0.00	0.20	0.00	**0.05**

EXT1	0.00	0.00	0.00	0.00	3.26	0.00	0.00	0.00	**0.25**
F11	0.00	0.00	0.00	0.00	12.40	0.00	0.00	0.00	**0.95**
F12	14.96	0.00	14.96	14.96	14.96	14.96	14.96	14.96	**9.96**
F2	0.00	0.00	0.00	234.43	0.00	0.00	0.00	0.00	**45.37**
F5	0.00	0.00	0.00	234.43	0.00	0.00	0.00	0.00	**45.37**
F5 [MBS]	762.07	487.29	225.19	495.10	653.08	157.60	337.90	1057.59	**514.61**
F8	2.05	4.28	2.05	13.88	31.97	2.05	10.91	6.09	**9.96**
F9	0.32	0.32	0.32	0.32	6.52	0.32	0.32	0.32	**0.80**
FAH	0.00	0.00	0.00	0.00	0.00	0.00	0.00	0.00	**0.00**
FANCC	0.00	54.46	0.00	0.00	0.00	0.00	15.92	0.00	**22.15**
FBN1	1.65	4.61	1.65	1.65	1.65	1.65	1.65	1.65	**2.64**
FCGR3A	0.00	0.00	0.00	0.00	0.00	0.00	2.01	0.00	**0.50**
FDFT1	0.00	0.00	0.00	0.00	0.00	0.00	0.00	0.00	**0.00**
FGFR1	4.32	8.46	4.32	4.32	4.32	4.32	12.85	8.37	**8.21**
FGFR2	4.32	8.32	4.32	4.32	4.32	4.32	12.85	8.37	**8.16**
FGFR3	4.40	11.74	4.40	10.05	23.97	4.40	12.73	9.96	**12.05**
FGFRL1	0.00	0.00	0.00	0.00	0.00	0.00	0.00	0.00	**0.00**
FIP1L1	0.00	0.45	0.00	0.00	0.00	0.00	0.00	0.00	**0.15**
FIP1L1/PDGFRA	3.89	5.65	3.89	3.89	3.89	3.89	7.11	3.89	**5.28**
FKRP	0.00	0.00	0.00	0.00	0.00	0.00	0.00	0.00	**0.00**
FLCN	0.00	0.89	0.00	0.00	0.00	0.00	0.00	1.01	**0.40**
FLNA	0.00	0.00	0.00	0.00	0.00	0.00	0.60	0.51	**0.20**
FLT3	0.00	0.00	0.00	0.00	0.00	0.00	0.60	9.61	**1.10**
FMR1	1.89	8.43	1.89	1.89	354.92	1.89	1.89	1.89	**31.01**
FMR1 [MBS]	252.99	258.75	225.19	126.28	99.17	128.57	291.36	33.37	**203.25**
FOXC2	0.00	0.00	0.00	0.00	0.00	0.00	0.00	0.51	**0.05**
FOXL2	0.00	0.00	0.00	0.00	0.00	0.00	0.00	0.00	**0.00**
FRAXA	0.00	0.00	0.00	0.00	0.00	0.00	0.00	160.26	**15.78**
FRAXE	0.00	0.00	0.00	0.00	0.00	0.00	0.00	160.26	**15.78**
FXN	1.26	4.17	1.26	33.67	7.14	1.26	53.67	13.90	**23.15**
FZD9	0.00	0.00	0.00	0.00	0.00	0.00	0.00	0.00	**0.00**
G6PC	0.00	8.33	0.00	0.00	0.00	0.00	0.00	0.00	**2.79**
GABRB3	0.00	0.00	0.00	0.00	1.30	0.00	0.00	0.00	**0.10**

GAK	0.00	0.00	0.00	0.00	0.00	0.00	0.00	0.00	**0.00**
GALT	0.00	0.00	0.00	6.43	1.30	0.00	0.00	1.52	**1.49**
GBA	1.40	1.40	1.40	1.40	0.00	1.40	1.40	1.40	**1.29**
GCH1	0.00	0.00	0.00	0.00	0.00	0.00	0.00	0.00	**0.00**
GCK	0.19	0.19	0.19	0.26	0.19	0.19	0.19	0.19	**0.20**
GDAP1	0.00	0.00	0.00	0.00	0.00	0.00	0.00	0.00	**0.00**
GFAP	0.00	0.00	0.00	0.00	0.00	0.00	0.00	1.01	**0.10**
GJB1	0.75	1.93	0.75	0.75	0.75	0.75	0.75	6.81	**1.74**
GJB2	12.50	50.29	12.50	35.90	69.26	12.50	94.70	33.73	**56.40**
GJB6	7.27	7.72	7.27	7.27	64.03	7.27	105.18	28.51	**38.03**
GLA	0.54	0.54	0.54	0.54	1.96	0.54	0.54	0.54	**0.65**
GLDC	0.00	0.00	0.00	0.00	0.00	0.00	0.00	0.00	**0.00**
GLUD1	0.00	0.00	0.00	0.00	0.00	0.00	0.00	0.00	**0.00**
GNB1	0.00	0.00	0.00	0.00	1.30	0.00	0.00	0.00	**0.10**
GP1BA	1.32	1.32	1.32	1.32	1.32	1.32	283.50	16.49	**72.53**
GRN	0.00	0.00	0.00	0.00	0.00	0.00	0.00	0.00	**0.00**
H19	2.58	2.58	2.58	2.58	2.58	2.58	4.23	2.58	**2.99**
HADHA	0.00	0.00	0.00	0.26	2.61	0.00	0.00	2.53	**0.50**
HBA1	21.43	20.58	21.43	21.43	249.79	21.43	175.00	586.12	**132.11**
HBA2	21.43	20.58	21.43	21.43	249.79	21.43	175.00	586.12	**132.11**
HBB	3.61	19.75	3.61	3.61	63.43	3.61	37.33	298.34	**50.92**
HBD	0.33	1.22	0.33	0.33	0.33	0.33	34.05	0.33	**8.96**
HBG1	0.00	1.79	0.00	0.00	0.00	0.00	0.00	0.00	**0.60**
HBG2	0.00	1.79	0.00	0.00	0.00	0.00	0.00	0.00	**0.60**
HEXA	3.87	69.32	3.87	3.87	3.87	3.87	125.04	3.87	**55.70**
HEXB	0.60	0.60	0.60	0.60	0.60	0.60	0.60	0.60	**0.60**
HFE	0.00	13.54	0.00	841.14	0.00	0.00	3.02	0.00	**168.07**
HFE [MBS]	2042.46	1088.12	450.37	1948.52	860.56	868.87	901.68	1472.64	**1232.87**
HIC1	0.00	0.00	0.00	0.00	1.30	0.00	0.00	0.00	**0.10**
HIPK3	0.00	0.00	0.00	0.00	0.00	0.00	0.00	0.00	**0.00**
HIRA	0.00	2.98	0.00	3.09	25.44	0.00	0.00	59.65	**9.41**
HLA-A	1.98	1.98	1.98	1.98	1.98	1.98	64.48	2479.13	**261.34**
HLA-B	1.32	38.37	1.32	1.32	1.32	1.32	40.30	2478.47	**267.26**

HLA-B [MBS]	3.09	2.23	15.01	1.03	72.42	14.52	108.20	0.00	**33.75**
HLA-C	0.33	0.33	0.33	0.33	0.33	0.33	16.12	2477.48	**248.15**
HLA-DPB1	0.00	0.00	0.00	0.00	0.00	0.00	4.03	2477.15	**244.91**
HLA-DQA1	19.36	0.66	19.36	19.36	19.36	19.36	79.15	19.36	**27.88**
HLA-DQB1	0.66	0.66	0.66	0.66	0.66	0.66	60.45	2477.81	**259.35**
HLA-DRB1	20.02	1.32	20.02	20.02	20.02	20.02	42.88	2497.17	**263.33**
HLA-DRB3	0.13	0.13	0.13	0.13	0.13	0.13	3.02	2477.28	**244.76**
HLA-DRB4	0.00	0.00	0.00	0.00	0.00	0.00	0.00	2477.15	**243.92**
HLA-DRB5	0.00	0.00	0.00	0.00	0.00	0.00	0.00	2477.15	**243.92**
HMBS	0.00	0.00	0.00	0.00	0.00	0.00	0.00	0.00	**0.00**
HMGCL	0.00	0.00	0.00	0.00	0.00	0.00	0.00	0.00	**0.00**
HMHA1	0.00	0.00	0.00	0.00	0.00	0.00	1.01	0.00	**0.25**
HNF1A	0.19	0.19	0.19	2.06	0.19	0.19	0.19	0.19	**0.55**
HNF4A	0.06	0.06	0.06	0.00	0.06	0.06	0.06	0.06	**0.05**
HPRT1	0.00	0.00	0.00	0.00	0.00	0.00	0.00	0.00	**0.00**
HRAS	0.00	0.00	0.00	0.00	0.00	0.00	0.00	0.00	**0.00**
HSD17B4	0.16	0.16	0.16	0.16	0.00	0.16	0.16	0.16	**0.15**
HTT	8.29	20.79	8.29	53.30	46.78	8.29	86.06	44.97	**46.94**
IDS	0.11	0.11	0.11	0.11	0.65	0.11	0.11	0.11	**0.15**
IDUA	0.27	0.27	0.27	0.27	0.00	0.27	0.27	0.27	**0.25**
IGH@	268.64	0.32	0.32	245.94	104.49	0.32	34.37	21.04	**70.59**
IGH@/BCL2	0.11	0.11	0.11	0.11	39.15	0.11	0.11	15.78	**4.63**
IGH@/CCND1	0.00	0.00	0.00	0.00	13.70	0.00	0.00	3.54	**1.39**
IGH@/MYC	0.00	0.00	0.00	0.00	0.00	0.00	0.00	2.02	**0.20**
IGHG1	0.00	1.04	0.00	0.00	0.00	0.00	0.00	16.18	**1.94**
IGHR	0.00	0.00	0.00	0.00	0.00	0.00	3.63	0.00	**0.90**
IKBKAP	0.00	54.46	0.00	0.00	0.00	0.00	15.92	0.00	**22.15**
IKBKG	0.70	0.70	0.70	0.70	13.05	0.70	0.70	0.70	**1.64**
IL2RG	0.00	0.30	0.00	0.00	0.00	0.00	0.00	0.00	**0.10**
IL7R	0.00	0.00	0.00	0.00	0.00	0.00	0.00	0.00	**0.00**
ITGA2	1.32	1.32	1.32	1.32	1.32	1.32	283.50	16.49	**72.53**
ITGA2B	1.32	1.32	1.32	1.32	1.32	1.32	357.45	16.49	**90.80**
ITGB3	1.32	1.32	1.32	1.32	1.32	1.32	359.46	16.49	**91.29**

ITGB4	0.00	0.00	0.00	2.83	0.00	0.00	0.00	0.00	**0.55**
JAG1	0.45	0.45	0.45	0.45	1.75	0.45	0.65	0.95	**0.65**
JAK2	3.11	57.71	3.11	3.11	166.93	3.11	48.75	38.49	**48.63**
KAL1	0.00	0.00	0.00	0.00	4.57	0.00	0.00	1.01	**0.45**
KCNE1	0.33	0.33	0.33	0.33	0.33	0.33	8.66	2.86	**2.64**
KCNE2	0.33	0.33	0.33	0.33	0.33	0.33	8.66	2.86	**2.64**
KCNH2	0.33	0.33	0.33	0.33	0.33	0.33	8.66	2.86	**2.64**
KCNJ11	0.19	0.19	0.19	0.00	0.19	0.19	0.19	0.19	**0.15**
KCNJ2	0.33	0.33	0.33	0.33	0.33	0.33	8.66	0.33	**2.39**
KCNJ6	0.00	0.00	0.00	0.00	0.00	0.00	0.00	0.00	**0.00**
KCNQ1	0.33	0.33	0.33	0.33	0.33	0.33	8.66	2.86	**2.64**
KCNQ1OT1	2.05	2.05	2.05	2.05	2.05	2.05	2.62	2.05	**2.19**
KEL	0.07	0.07	0.07	0.07	5.29	0.07	7.05	2.59	**2.44**
KIR3DL1	0.20	0.20	0.20	0.20	0.20	0.20	6.04	0.20	**1.64**
KIT	3.70	3.85	3.70	3.70	3.70	3.70	21.96	3.70	**8.26**
KMS	0.00	0.00	0.00	0.00	0.65	0.00	0.00	0.00	**0.05**
KRAS	0.00	0.00	0.00	0.00	0.00	0.00	0.00	0.00	**0.00**
KRIT1	0.00	0.00	0.00	0.00	0.00	0.00	0.00	0.00	**0.00**
KRT10	0.00	0.00	0.00	0.00	0.00	0.00	0.00	0.00	**0.00**
KRT14	0.00	0.00	0.00	4.89	0.00	0.00	0.00	0.00	**0.95**
KRT5	0.00	0.00	0.00	4.89	0.00	0.00	0.00	0.00	**0.95**
L1CAM	0.00	0.00	0.00	0.00	0.00	0.00	1.21	2.02	**0.50**
LAMA3	0.00	0.00	0.00	2.83	0.00	0.00	0.00	0.00	**0.55**
LAMB3	0.00	0.00	0.00	2.83	0.00	0.00	0.00	0.00	**0.55**
LAMC2	0.00	0.00	0.00	2.83	0.00	0.00	0.00	0.00	**0.55**
LDLR	0.00	0.00	0.00	0.00	0.00	0.00	0.00	0.00	**0.00**
LETM1	0.00	0.00	0.00	0.00	0.00	0.00	0.00	0.00	**0.00**
LIMK1	0.00	0.00	0.00	0.00	11.09	0.00	0.00	7.58	**1.59**
LLGL1	0.00	0.00	0.00	0.00	1.30	0.00	0.00	0.00	**0.10**
LMNA	0.94	1.39	0.94	0.94	0.94	0.94	10.22	0.94	**3.38**
LRRK2	0.13	0.13	0.13	0.13	0.13	0.13	0.20	0.13	**0.15**
MAPT	0.00	0.00	0.00	0.00	0.00	0.00	0.20	3.03	**0.35**
MAT1A	0.00	0.00	0.00	0.00	0.00	0.00	0.00	0.00	**0.00**

MBP	0.00	0.00	0.00	0.00	0.00	0.00	0.00	0.00	**0.00**
MC2R	0.19	0.19	0.19	0.00	0.19	0.19	0.19	0.19	**0.15**
MC4R	0.06	0.06	0.06	0.00	0.06	0.06	0.06	0.06	**0.05**
MCOLN1	0.00	8.33	0.00	0.00	0.00	0.00	0.00	0.00	**2.79**
MECP2	4.76	8.69	4.76	4.76	4.76	4.76	4.76	35.57	**9.11**
MEFV	7.44	24.66	7.44	7.44	15.16	7.44	7.44	10.47	**14.09**
MEN1	0.66	0.66	0.66	6.06	1.97	0.66	5.04	13.30	**4.13**
MET	0.05	0.35	0.05	0.05	1.30	0.05	0.05	0.05	**0.25**
MFAP4	0.00	0.00	0.00	0.00	1.30	0.00	0.00	0.00	**0.10**
MFN2	0.45	5.21	0.45	0.45	0.45	0.45	0.45	5.00	**2.49**
MGMT	0.00	0.00	0.00	0.00	0.00	0.00	0.00	0.00	**0.00**
MKRN3	0.00	0.00	0.00	0.00	1.30	0.00	0.00	0.00	**0.10**
MLH1	6.40	15.18	6.40	21.83	23.37	6.40	25.63	59.99	**23.64**
MLL	0.49	1.53	0.49	0.49	12.40	0.49	0.49	0.49	**1.74**
MLL/AFF1	0.00	0.00	0.00	30.86	0.00	0.00	0.00	0.00	**5.97**
MNX1	0.00	0.00	0.00	0.00	0.00	0.00	0.00	0.00	**0.00**
MPZ	0.22	1.64	0.22	0.22	0.22	0.22	0.22	3.26	**1.00**
MSH2	6.34	13.63	6.34	27.17	23.69	6.34	23.28	59.92	**23.60**
MSH6	3.55	5.78	3.55	3.55	7.35	3.55	19.05	56.63	**13.64**
MSI#	0.60	22.91	0.60	0.60	119.99	0.60	5.24	0.60	**18.32**
MSRA	0.00	0.00	0.00	0.00	0.00	0.00	0.00	0.00	**0.00**
MT-ATP6	3.62	3.62	3.62	57.38	4.87	3.62	34.31	3.62	**21.70**
MT-CO1	0.07	0.07	0.07	0.07	0.07	0.07	0.00	0.07	**0.05**
MT-CO2	0.07	0.07	0.07	0.07	0.07	0.07	0.00	0.07	**0.05**
MT-CO3	0.07	0.07	0.07	0.07	0.07	0.07	0.00	0.07	**0.05**
MT-CYB	0.00	0.00	0.00	6.17	0.00	0.00	0.00	0.00	**1.19**
MT-deletion#	0.13	0.13	0.13	21.74	0.13	0.13	7.05	0.13	**6.02**
MTHFR	0.00	0.00	0.00	67.51	0.00	0.00	6.04	0.00	**14.56**
MTM1	0.17	0.17	0.17	0.17	0.17	0.17	0.17	0.00	**0.15**
MT-ND1	0.65	0.65	0.65	6.82	7.12	0.65	6.50	0.65	**3.78**
MT-ND4	0.65	0.65	0.65	60.58	7.12	0.65	6.50	0.65	**14.19**
MT-ND5	0.26	1.74	0.26	0.26	11.89	0.26	0.46	0.26	**1.69**
MT-ND6	0.65	0.65	0.65	0.65	7.12	0.65	6.50	0.65	**2.59**

MT-RNR1	0.07	0.07	0.07	0.07	1.37	0.07	0.00	0.57	**0.20**
MT-TK	4.10	5.59	4.10	57.86	7.31	4.10	46.95	4.10	**25.84**
MT-TL1	0.31	0.31	0.31	54.06	15.86	0.31	19.85	0.31	**16.73**
MUTYH	1.75	6.29	1.75	1.75	108.27	1.75	16.87	1.75	**15.13**
MYBPC3	0.00	0.00	0.00	0.00	0.00	0.00	0.81	0.00	**0.20**
MYC	0.00	2.68	0.00	0.00	9.13	0.00	0.00	2.53	**1.84**
MYCN	0.00	0.00	0.00	11.06	0.00	0.00	0.40	0.00	**2.24**
MYH7	0.00	0.00	0.00	0.00	0.00	0.00	0.60	0.00	**0.15**
MYOT	0.00	0.00	0.00	0.00	0.00	0.00	0.00	0.00	**0.00**
NAGLU	0.05	0.05	0.05	0.05	0.00	0.05	0.05	0.05	**0.05**
NDN	0.00	0.00	0.00	0.00	1.30	0.00	0.00	0.00	**0.10**
NDP	0.00	0.00	0.00	1.54	0.00	0.00	0.00	0.00	**0.30**
NF1	0.00	0.00	0.00	0.00	0.00	0.00	0.00	6.57	**0.65**
NF2	0.59	0.59	0.59	0.59	0.00	0.59	3.82	3.12	**1.59**
NIPA1	0.07	0.15	0.07	0.07	0.07	0.07	0.07	0.07	**0.10**
NOD2	0.00	0.40	0.40	0.40	0.40	0.40	10.48	0.40	**2.89**
NOG	0.00	0.00	0.00	0.00	0.00	0.00	0.00	0.00	**0.00**
NOTCH3	4.04	4.04	4.04	2.89	4.04	4.04	4.04	18.35	**5.23**
NPC1	0.16	8.49	0.16	0.16	0.65	0.16	0.16	0.16	**2.99**
NR0B1	0.00	0.00	0.00	0.00	0.00	0.00	0.00	0.00	**0.00**
NR4A2	0.00	0.00	0.00	0.00	0.00	0.00	0.00	0.00	**0.00**
NSD1	0.00	0.00	0.00	0.00	1.30	0.00	0.00	3.03	**0.40**
OCRL	0.00	0.00	0.00	0.00	0.00	0.00	0.00	0.00	**0.00**
OTC	0.43	2.29	0.43	0.94	1.68	0.43	1.44	0.43	**1.49**
OTOF	0.00	0.00	0.00	0.00	0.00	0.00	0.00	0.00	**0.00**
PABPN1	0.00	0.00	0.00	5.40	0.00	0.00	0.00	0.00	**1.05**
PAFAH1B1	0.00	0.15	0.00	0.00	3.26	0.00	0.00	3.03	**0.60**
PAH	0.00	0.00	0.00	0.26	0.00	0.00	0.00	0.00	**0.05**
PALB2	0.00	0.00	0.00	0.00	0.00	0.00	0.00	0.00	**0.00**
PANK2	0.00	0.00	0.00	0.00	0.00	0.00	0.00	0.00	**0.00**
PARK2	0.26	0.26	0.26	0.26	0.26	0.26	1.01	0.26	**0.45**
PARK7	0.07	0.07	0.07	0.07	0.07	0.07	0.00	0.07	**0.05**
PAX6	0.00	0.00	0.00	0.00	3.91	0.00	0.00	0.00	**0.30**

PAX7/FOXO1A	0.00	0.00	0.00	0.00	0.00	0.00	0.60	0.00	**0.15**
PDCD6	0.00	0.00	0.00	0.00	0.00	0.00	0.00	0.00	**0.00**
PDHA1	0.00	0.00	0.00	0.00	0.00	0.00	0.20	0.00	**0.05**
PEX1	0.16	0.16	0.16	0.16	3.26	0.16	0.16	0.16	**0.40**
PEX7	0.16	0.16	0.16	0.16	0.00	0.16	0.16	0.16	**0.15**
PINK1	0.13	0.13	0.13	0.13	0.13	0.13	0.00	0.13	**0.10**
PITX2	0.00	0.00	0.00	0.00	0.00	0.00	0.00	0.00	**0.00**
PKP2	0.00	0.00	0.00	0.00	0.00	0.00	0.40	0.00	**0.10**
PLA2G6	0.00	0.00	0.00	0.00	0.00	0.00	0.00	0.00	**0.00**
PML/RARA	0.00	2.53	0.00	0.00	3.91	0.00	10.48	18.20	**5.53**
PMM2	0.43	0.43	0.43	0.43	0.00	0.43	0.43	0.43	**0.40**
PMP22	3.22	11.90	3.22	3.22	24.09	3.22	26.39	18.38	**14.93**
PMS2	1.02	1.02	1.02	1.02	4.57	1.02	1.83	2.04	**1.59**
POLG	0.20	0.20	0.20	0.20	0.20	0.20	0.60	0.20	**0.30**
POU1F1	0.00	0.00	0.00	0.00	0.00	0.00	0.00	0.00	**0.00**
PPOX	0.00	0.00	0.00	0.00	0.00	0.00	0.00	0.00	**0.00**
PPP2R2B	0.00	0.00	0.00	0.00	0.00	0.00	0.00	0.00	**0.00**
PPT1	0.16	0.16	0.16	0.16	0.00	0.16	0.16	0.16	**0.15**
PRF1	0.00	0.00	0.00	0.00	0.00	0.00	0.00	0.00	**0.00**
PRNP	2.05	2.05	2.05	2.05	2.05	2.05	4.43	3.06	**2.74**
PROP1	0.00	0.00	0.00	0.00	0.00	0.00	0.00	0.00	**0.00**
PRPSAP2	0.00	0.00	0.00	0.00	1.30	0.00	0.00	0.00	**0.10**
PRSS1	0.00	0.00	0.00	0.00	0.00	0.00	0.00	2.02	**0.20**
PSEN1	0.66	0.66	0.66	0.66	0.66	0.66	6.04	0.66	**1.99**
PSEN2	0.00	0.00	0.00	0.00	0.00	0.00	0.00	0.00	**0.00**
PTEN	0.15	1.79	0.15	0.15	3.41	0.15	0.15	2.17	**1.14**
PTPN11	0.00	0.00	0.00	0.00	0.00	0.00	0.00	8.59	**0.85**
PYGM	0.06	0.06	0.06	0.06	0.06	0.06	0.06	0.51	**0.10**
RAF1	0.00	0.00	0.00	0.00	0.00	0.00	0.00	0.00	**0.00**
RAG1	0.00	0.00	0.00	0.00	0.00	0.00	0.00	0.00	**0.00**
RAG2	0.00	0.00	0.00	0.00	0.00	0.00	0.00	0.00	**0.00**
RAI1	0.00	0.00	0.00	0.00	16.31	0.00	0.00	4.04	**1.64**
RARA	0.00	0.00	0.00	0.00	5.87	0.00	0.00	0.00	**0.45**

RB1	1.76	1.76	1.76	1.76	58.53	1.76	11.37	5.80	**8.86**
RET	1.18	6.02	1.18	1.18	1.84	1.18	16.04	3.71	**6.77**
RHCE	0.07	0.07	0.07	0.07	5.29	0.07	7.05	0.07	**2.19**
RHD	0.51	3.04	0.51	0.51	5.73	0.51	7.50	0.51	**3.48**
RUNX1/RUNX1T1	0.00	0.00	0.00	0.00	4.57	0.00	2.42	5.56	**1.49**
RYR1	0.72	0.72	0.72	0.72	0.72	0.72	0.72	2.02	**0.85**
SAMD12	0.00	0.00	0.00	0.00	0.00	0.00	0.00	0.00	**0.00**
SBDS	0.00	0.00	0.00	0.00	0.00	0.00	0.00	1.01	**0.10**
SCN1A	0.20	0.20	0.20	0.20	0.20	0.20	1.21	0.20	**0.45**
SCN4A	0.06	0.06	0.06	1.60	0.06	0.06	0.06	1.52	**0.50**
SCN5A	0.33	0.33	0.33	0.33	0.33	0.33	8.66	0.33	**2.39**
SCNN1D	0.00	0.00	0.00	0.00	1.30	0.00	0.00	0.00	**0.10**
SDHB	4.41	5.51	4.41	4.41	4.41	4.41	4.41	4.41	**4.78**
SDHD	2.77	2.83	2.77	2.77	2.77	2.77	2.77	2.77	**2.79**
SEMA5A	0.00	0.00	0.00	0.00	0.00	0.00	0.00	0.00	**0.00**
sent o'seas#	68.21	1.07	0.33	21.42	41.43	0.33	50.17	0.33	**21.21**
SEPN1	0.00	0.00	0.00	0.00	0.00	0.00	0.40	0.00	**0.10**
SERPINA1	0.00	0.00	0.00	125.25	0.00	0.00	1.21	401.91	**64.12**
SERPING1	2.24	0.00	2.24	2.24	2.24	2.24	2.24	2.24	**1.49**
SFTPB	0.00	0.60	0.00	0.00	0.00	0.00	0.00	0.00	**0.20**
SGSH	0.43	0.43	0.43	0.43	0.00	0.43	0.43	0.43	**0.40**
SH2D1A	0.00	0.00	0.00	0.00	0.00	0.00	0.00	1.01	**0.10**
SHOX	0.00	0.00	0.00	0.00	11.74	0.00	0.00	0.00	**0.90**
SKI	0.00	0.00	0.00	0.00	1.30	0.00	0.00	0.00	**0.10**
SLBP	0.00	0.00	0.00	0.00	0.00	0.00	0.00	0.00	**0.00**
SLC14A1	0.07	0.07	0.07	0.07	5.29	0.07	7.05	2.59	**2.44**
SLC26A4	0.00	0.00	0.00	0.00	0.00	0.00	0.00	20.73	**2.04**
SMAD4	0.07	0.15	0.07	0.07	0.07	0.07	0.07	0.07	**0.10**
SMARCB1	0.00	0.00	0.00	0.00	0.00	0.00	0.40	0.00	**0.10**
SMCR	0.00	0.15	0.00	0.00	0.00	0.00	0.00	0.00	**0.05**
SMN1	9.27	17.60	9.27	9.27	19.60	9.27	92.48	22.42	**34.70**
SMPD1	0.00	0.00	0.00	0.00	0.00	0.00	15.92	0.00	**3.93**
SNAP29	0.00	0.00	0.00	0.00	1.30	0.00	0.00	0.00	**0.10**

SNCA	0.35	0.50	0.35	0.35	0.35	0.35	0.15	0.35	**0.35**
SNCAIP	0.00	0.00	0.00	0.00	0.00	0.00	0.00	0.00	**0.00**
SNRPN	0.49	11.58	0.49	17.21	30.24	0.49	36.36	48.52	**23.30**
SOD1	1.00	2.41	1.00	1.00	1.00	1.00	1.21	2.70	**1.69**
Somatic hypermutation#	0.00	0.00	0.00	0.00	0.65	0.00	0.00	0.00	**0.05**
SOS1	0.00	0.00	0.00	0.00	0.00	0.00	0.00	0.00	**0.00**
SPAST	0.07	0.15	0.07	0.07	0.07	0.07	0.07	5.64	**0.65**
SPG3A	0.07	0.15	0.07	0.07	0.07	0.07	0.07	0.07	**0.10**
SPON2	0.00	0.00	0.00	0.00	0.00	0.00	0.00	0.00	**0.00**
SPTLC1	0.45	1.93	0.45	0.45	0.45	0.45	0.45	0.45	**0.95**
SRD5A2	0.00	0.00	0.00	0.00	0.00	0.00	0.00	0.00	**0.00**
SRY	0.45	2.98	0.45	86.35	1.75	0.45	0.45	2.47	**18.22**
SS18	0.00	0.00	0.00	0.00	1.30	0.00	0.00	0.00	**0.10**
STK11	0.97	4.31	0.97	0.97	0.97	0.97	0.97	0.97	**2.09**
STR#	3.94	7.89	3.94	9.35	32.94	3.94	6.13	3.94	**9.06**
STS	0.00	0.00	0.00	0.00	3.26	0.00	0.00	0.00	**0.25**
STX1A	0.00	0.00	0.00	0.00	1.30	0.00	0.00	0.00	**0.10**
Subtel deletion#	0.00	19.49	0.00	0.00	488.67	0.00	101.55	33.37	**72.18**
SURF1	0.13	0.13	0.13	0.13	0.13	0.13	1.21	0.13	**0.40**
TACC3	0.00	0.00	0.00	0.00	0.00	0.00	0.00	0.00	**0.00**
TBP	0.00	0.74	0.00	0.00	0.00	0.00	0.00	0.00	**0.25**
TCF3	0.49	0.49	0.49	0.49	5.22	0.49	0.49	0.49	**0.85**
TCF3/PBX1	0.00	0.00	0.00	30.86	0.00	0.00	0.00	0.00	**5.97**
TERT	0.00	0.00	0.00	0.00	0.00	0.00	0.00	0.00	**0.00**
TGFBR1	0.22	0.30	0.22	0.22	0.22	0.22	0.22	0.22	**0.25**
TGFBR2	0.22	0.30	0.22	0.22	0.22	0.22	0.22	0.22	**0.25**
TH	0.00	0.00	0.00	0.00	0.00	0.00	0.00	0.00	**0.00**
TNFRSF13B	0.00	0.00	0.00	0.00	1.30	0.00	0.00	0.00	**0.10**
TNFRSF18	0.00	0.00	0.00	0.00	1.30	0.00	0.00	0.00	**0.10**
TNFRSF1A	3.29	8.63	3.29	3.29	3.29	3.29	3.29	3.29	**5.08**
TNFRSF4	0.00	0.00	0.00	0.00	1.30	0.00	0.00	0.00	**0.10**
TOR1A	1.09	1.09	1.09	1.09	5.88	1.09	1.09	3.46	**1.69**
TP53	0.79	1.83	0.79	0.79	24.28	0.79	3.83	1.30	**3.73**

TP73	0.00	0.00	0.00	0.00	1.30	0.00	0.00	0.00	**0.10**
TPM3	0.11	0.11	0.11	0.11	0.11	0.11	0.11	0.51	**0.15**
TPMT	0.05	0.05	0.05	0.05	26.75	0.05	0.05	39.99	**6.02**
TPP1	0.59	0.59	0.59	0.59	5.22	0.59	0.59	0.59	**0.95**
TRAPPC2	0.11	0.11	0.11	0.11	0.00	0.11	0.11	0.11	**0.10**
TRB@	0.00	0.00	0.00	245.62	0.00	0.00	42.11	0.00	**57.94**
TRG@	0.16	0.16	0.16	245.78	65.90	0.16	55.77	58.80	**72.23**
TRPS1	0.00	0.00	0.00	0.00	3.26	0.00	0.00	0.00	**0.25**
TRPV1	0.00	0.00	0.00	0.00	1.30	0.00	0.00	0.00	**0.10**
TSC1	0.22	0.15	0.22	0.22	0.22	0.22	0.22	4.77	**0.65**
TSC2	0.22	0.15	0.22	0.22	0.22	0.22	1.03	4.77	**0.85**
TTN	0.00	0.00	0.00	0.00	0.00	0.00	0.00	0.00	**0.00**
TTR	0.00	0.30	0.00	0.00	0.00	0.00	0.60	0.00	**0.25**
TWIST1	4.19	8.18	4.19	4.19	5.49	4.19	4.19	4.19	**5.63**
TWISTNB	0.00	0.00	0.00	0.00	1.30	0.00	0.00	0.00	**0.10**
TYR	0.00	0.00	0.00	0.00	0.00	0.00	0.00	0.51	**0.05**
UBE3A	0.37	8.18	0.37	3.72	29.73	0.37	36.24	35.26	**18.17**
UCHL1	0.00	0.00	0.00	0.00	0.00	0.00	0.00	0.00	**0.00**
UGT1A1	0.17	0.17	0.17	0.17	0.17	0.17	16.29	46.00	**8.66**
UNC13D	0.00	0.00	0.00	0.00	0.00	0.00	0.00	1.52	**0.15**
VHL	3.79	4.75	3.79	3.79	7.95	3.79	3.79	8.34	**4.88**
VKORC1	2.31	2.31	2.31	2.31	2.31	2.31	12.09	2.31	**4.73**
VWF	0.00	0.00	0.00	0.00	3.26	0.00	0.00	0.00	**0.25**
WAS	0.00	0.00	0.00	0.00	0.00	0.00	0.00	1.52	**0.15**
WFS1	0.15	0.60	0.15	0.15	0.15	0.15	0.15	0.15	**0.30**
WHSC1	0.00	0.00	0.00	0.00	1.96	0.00	0.00	0.00	**0.15**
WHSC2	0.00	0.00	0.00	0.00	0.00	0.00	0.00	0.00	**0.00**
WT1	0.73	0.73	0.73	0.73	0.73	0.73	0.40	0.73	**0.65**
YWHAE	0.00	0.00	0.00	0.00	1.96	0.00	0.00	1.52	**0.30**

15.2.5 RATE OF TESTING FOR INTRA- VERSUS INTER-STATE SAMPLES

Type of test HGNC test name, or non-standard name (indicated by #).
Popl'n in testing region combined population in all regions offering the type of test in 2006.

Assays in testing region actual number of assays in all regions offering the type of test in 2006.
Rate in testing region number of assays per person in all regions offering the type of test in 2006.
Popl'n outside testing region combined population in all regions not offering the type of test in 2006.
Assays outside testing region actual number of assays done on samples from patients in regions not offering the type of test in 2006.
Expected assays outside region expected number of assays on samples from patients in regions not offering the type of test if the rate of testing had been the same as *Rate in testing region*.
p-value probability that the difference between actual and observed numbers of assays is due to chance (Poisson probability mass function)

Type of test	Popl'n in testing region	Assays in testing region	Rate in testing region	Popl'n outside testing region	Assays outside testing region	Expected assays outside region	p-value
ABCC8	-	0		20,567,429	4		n/a
ABCD1	1,550,750	8	5.2E-06	19,016,679	22	98.1	0.000000
ABL1	2,030,643	8	3.9E-06	18,536,786	0	73.0	0.000000
ACADS	-	0		20,567,429	0		n/a
ACADVL	-	0		20,567,429	0		n/a
ACTA1	2,030,643	2	9.8E-07	18,536,786	28	18.3	0.008072
ADA	-	0		20,567,429	0		n/a
ADSL	-	0		20,567,429	0		n/a
AFF2	1,550,750	6	3.9E-06	19,016,679	4	73.6	0.000000
ALDOB	12,922,143	5	3.9E-07	7,645,286	6	3.0	0.048318
ALK	3,581,393	20	5.6E-06	16,986,036	0	94.9	0.000000
ALPL	-	0		20,567,429	0		n/a
APC	19,547,286	290	1.5E-05	1,020,143	65	15.1	0.000000
APOB	3,581,393	26	7.3E-06	16,986,036	0	123.3	0.000000
APOE	19,547,286	1362	7.0E-05	1,020,143	0	71.1	0.000000
APP	5,074,393	31	6.1E-06	15,493,036	10	94.6	0.000000
AR	19,547,286	124	6.3E-06	1,020,143	17	6.5	0.000266
ARSA	1,550,750	5	3.2E-06	19,016,679	6	61.3	0.000000
ARSE	-	0		20,567,429	0		n/a
ARVCF	1,550,750	2	1.3E-06	19,016,679	0	24.5	0.000000
ARX	3,581,393	20	5.6E-06	16,986,036	2	94.9	0.000000
ASPA	11,937,786	449	3.8E-05	8,629,643	0	324.6	0.000000
ATM	10,444,786	43	4.1E-06	10,122,643	0	41.7	0.000000

ATN1	19,547,286	185	9.5E-06	1,020,143	2	9.7	0.002988
ATP13A2	-	0		20,567,429	0		n/a
ATP1A2	-	0		20,567,429	0		n/a
ATP7A	5,074,393	12	2.4E-06	15,493,036	0	36.6	0.000000
ATRX	-	0		20,567,429	0		n/a
ATXN1	19,547,286	394	2.0E-05	1,020,143	4	20.6	0.000009
ATXN2	19,547,286	398	2.0E-05	1,020,143	4	20.8	0.000007
ATXN3	19,547,286	398	2.0E-05	1,020,143	4	20.8	0.000007
ATXN7	19,547,286	401	2.1E-05	1,020,143	4	20.9	0.000007
AVP	-	0		20,567,429	0		n/a
AZF#	8,894,036	102	1.1E-05	11,673,393	0	133.9	0.000000
BCHE	6,058,750	37	6.1E-06	14,508,679	0	88.6	0.000000
BCL2	17,847,322	1121	6.3E-05	2,720,107	0	170.9	0.000000
BCL6	2,030,643	13	6.4E-06	18,536,786	0	118.7	0.000000
BCR/ABL1	19,547,286	1623	8.3E-05	1,020,143	1147	84.7	0.000000
BDNF	-	0		20,567,429	0		n/a
BLM	11,937,786	136	1.1E-05	8,629,643	0	98.3	0.000000
BMPR1A	6,863,393	3	4.4E-07	13,704,036	3	6.0	0.089679
BRAF	13,488,536	108	8.0E-06	7,078,893	2	56.7	0.000000
BRCA1	19,547,286	1003	5.1E-05	1,020,143	190	52.3	0.000000
BRCA2	19,547,286	957	4.9E-05	1,020,143	207	49.9	0.000000
BTK	2,030,643	2	9.8E-07	18,536,786	0	18.3	0.000000
CACNA1A	19,547,286	407	2.1E-05	1,020,143	26	21.2	0.047430
CACNA1S	6,058,750	12	2.0E-06	14,508,679	2	28.7	0.000000
CASR	6,058,750	20	3.3E-06	14,508,679	2	47.9	0.000000
CAV3	2,030,643	1	4.9E-07	18,536,786	0	9.1	0.000109
CBFB	8,414,143	12	1.4E-06	12,153,286	0	17.3	0.000000
CBFB/MYH11	7,105,036	20	2.8E-06	13,462,393	0	37.9	0.000000
CCND1	11,133,143	577	5.2E-05	9,434,286	0	489.0	0.000000
CCND1/IGHG1	10,444,786	111	1.1E-05	10,122,643	0	107.6	0.000000
CCR5	2,030,643	44	2.2E-05	18,536,786	0	401.7	0.000000
CD109	7,105,036	1170	1.6E-04	13,462,393	20	2216.9	0.000000
CD177	5,074,393	10	2.0E-06	15,493,036	0	30.5	0.000000

CD2	-	0		20,567,429	0		n/a
CD40LG	-	0		20,567,429	0		n/a
CDC2L1	3,581,393	23	6.4E-06	16,986,036	0	109.1	0.000000
CDC45L	1,550,750	2	1.3E-06	19,016,679	0	24.5	0.000000
CDC73	2,030,643	1	4.9E-07	18,536,786	0	9.1	0.000109
CDH1	6,863,393	6	8.7E-07	13,704,036	5	12.0	0.012889
CDKN1B	1,550,750	3	1.9E-06	19,016,679	0	36.8	0.000000
CDKN1C	-	0		20,567,429	0		n/a
CDKN2A	6,863,393	1	1.5E-07	13,704,036	2	2.0	0.270670
CFTR	19,877,965	10773	5.4E-04	689,464	323	373.7	0.000606
CHD7	2,030,643	1	4.9E-07	18,536,786	0	9.1	0.000109
Chimerism#	10,444,786	46	4.4E-06	10,122,643	0	44.6	0.000000
CLCN1	2,030,643	6	3.0E-06	18,536,786	15	54.8	0.000000
CLDN5	1,550,750	2	1.3E-06	19,016,679	0	24.5	0.000000
CLIP2	1,550,750	2	1.3E-06	19,016,679	0	24.5	0.000000
CLN3	1,550,750	2	1.3E-06	19,016,679	25	24.5	0.079162
CLTCL1	1,550,750	2	1.3E-06	19,016,679	0	24.5	0.000000
COL2A1	5,074,393	3	5.9E-07	15,493,036	0	9.2	0.000105
COL3A1	6,863,393	4	5.8E-07	13,704,036	0	8.0	0.000340
COL7A1	4,028,107	9	2.2E-06	16,539,322	0	37.0	0.000000
CPOX	-	0		20,567,429	0		n/a
CPS1	-	0		20,567,429	0		n/a
CREBBP	2,030,643	1	4.9E-07	18,536,786	7	9.1	0.113738
CRK	3,581,393	6	1.7E-06	16,986,036	0	28.5	0.000000
CTBP1	-	0		20,567,429	0		n/a
CTLA4	5,074,393	20	3.9E-06	15,493,036	0	61.1	0.000000
CTNND2	-	0		20,567,429	0		n/a
CTNS	1,550,750	3	1.9E-06	19,016,679	4	36.8	0.000000
CTSB	-	0		20,567,429	0		n/a
CYBB	2,030,643	1	4.9E-07	18,536,786	0	9.1	0.000109
CYP21A2	6,058,750	23	3.8E-06	14,508,679	29	55.1	0.000042
CYP2C19	5,074,393	100	2.0E-05	15,493,036	40	305.3	0.000000
CYP2C9	5,074,393	60	1.2E-05	15,493,036	35	183.2	0.000000

CYP2D6	5,074,393	100	2.0E-05	15,493,036	40	305.3	0.000000
D13S319#	-	0		20,567,429	0		n/a
D19S545#	2,030,643	32	1.6E-05	18,536,786	0	292.1	0.000000
D19S851#	2,030,643	32	1.6E-05	18,536,786	0	292.1	0.000000
D4Z4#	8,894,036	90	1.0E-05	11,673,393	49	118.1	0.000000
D5S721#	1,550,750	1	6.4E-07	19,016,679	0	12.3	0.000005
D7S613#	3,581,393	30	8.4E-06	16,986,036	0	142.3	0.000000
DARC	8,655,786	48	5.5E-06	11,911,643	1	66.1	0.000000
DCX	2,030,643	2	9.8E-07	18,536,786	11	18.3	0.022164
DDIT3	1,550,750	3	1.9E-06	19,016,679	0	36.8	0.000000
DGCR	1,550,750	5	3.2E-06	19,016,679	0	61.3	0.000000
DGCR2	2,030,643	5	2.5E-06	18,536,786	0	45.6	0.000000
DHCR7	-	0		20,567,429	6		n/a
DMD	19,547,286	435	2.2E-05	1,020,143	80	22.7	0.000000
DMPK	19,547,286	574	2.9E-05	1,020,143	48	30.0	0.000586
DYRK1A	-	0		20,567,429	0		n/a
EBP	1,550,750	2	1.3E-06	19,016,679	0	24.5	0.000000
EDA	-	0		20,567,429	0		n/a
EGFR	8,655,786	180	2.1E-05	11,911,643	56	247.7	0.000000
ELN	10,444,786	33	3.2E-06	10,122,643	0	32.0	0.000000
EMD	5,074,393	28	5.5E-06	15,493,036	12	85.5	0.000000
ERBB2	-	0		20,567,429	0		n/a
ETV6/RUNX1	12,683,893	158	1.2E-05	7,883,536	9	98.2	0.000000
EWSR1	8,414,143	12	1.4E-06	12,153,286	0	17.3	0.000000
EWSR1/FLI1	5,074,393	3	5.9E-07	15,493,036	0	9.2	0.000105
EWSR1/WT1	5,074,393	1	2.0E-07	15,493,036	0	3.1	0.047209
EXT1	1,550,750	5	3.2E-06	19,016,679	0	61.3	0.000000
F11	1,550,750	19	1.2E-05	19,016,679	0	233.0	0.000000
F12	-	0		20,567,429	200		n/a
F2	4,028,107	912	2.3E-04	16,539,322	0	3742.6	0.000000
F5	4,028,107	912	2.3E-04	16,539,322	0	3742.6	0.000000
F8	19,547,286	162	8.3E-06	1,020,143	38	8.5	0.000000
F9	1,550,750	10	6.4E-06	19,016,679	6	122.6	0.000000

FAH	-	0		20,567,429	0		n/a
FANCC	11,937,786	445	3.7E-05	8,629,643	0	321.7	0.000000
FBN1	6,863,393	31	4.5E-06	13,704,036	22	61.9	0.000000
FCGR3A	5,074,393	10	2.0E-06	15,493,036	0	30.5	0.000000
FDFT1	-	0		20,567,429	0		n/a
FGFR1	13,968,429	107	7.7E-06	6,599,000	58	50.5	0.030976
FGFR2	13,968,429	106	7.6E-06	6,599,000	58	50.1	0.028818
FGFR3	19,547,286	183	9.4E-06	1,020,143	59	9.6	0.000000
FGFRL1	-	0		20,567,429	0		n/a
FIP1L1	6,863,393	3	4.4E-07	13,704,036	0	6.0	0.002504
FIP1L1/PDGFRA	11,937,786	54	4.5E-06	8,629,643	52	39.0	0.007878
FKRP	-	0		20,567,429	0		n/a
FLCN	8,894,036	8	9.0E-07	11,673,393	0	10.5	0.000028
FLNA	7,105,036	4	5.6E-07	13,462,393	0	7.6	0.000511
FLT3	7,105,036	22	3.1E-06	13,462,393	0	41.7	0.000000
FMR1	8,414,143	588	7.0E-05	12,153,286	35	849.3	0.000000
FOXC2	2,030,643	1	4.9E-07	18,536,786	0	9.1	0.000109
FOXL2	-	0		20,567,429	0		n/a
FRAXA	2,030,643	317	1.6E-04	18,536,786	0	2893.7	0.000000
FRAXE	2,030,643	317	1.6E-04	18,536,786	0	2893.7	0.000000
FXN	19,547,286	446	2.3E-05	1,020,143	19	23.3	0.059913
FZD9	-	0		20,567,429	0		n/a
G6PC	6,863,393	56	8.2E-06	13,704,036	0	111.8	0.000000
GABRB3	1,550,750	2	1.3E-06	19,016,679	0	24.5	0.000000
GAK	-	0		20,567,429	0		n/a
GALT	7,609,500	30	3.9E-06	12,957,929	0	51.1	0.000000
GBA	-	0		20,567,429	26		n/a
GCH1	-	0		20,567,429	0		n/a
GCK	4,028,107	1	2.5E-07	16,539,322	3	4.1	0.190062
GDAP1	-	0		20,567,429	0		n/a
GFAP	2,030,643	2	9.8E-07	18,536,786	0	18.3	0.000000
GJB1	8,894,036	25	2.8E-06	11,673,393	10	32.8	0.000002
GJB2	19,547,286	944	4.8E-05	1,020,143	189	49.3	0.000000

GJB6	15,519,179	654	4.2E-05	5,048,250	110	212.7	0.000000
GLA	1,550,750	3	1.9E-06	19,016,679	10	36.8	0.000000
GLDC	-	0		20,567,429	0		n/a
GLUD1	-	0		20,567,429	0		n/a
GNB1	1,550,750	2	1.3E-06	19,016,679	0	24.5	0.000000
GP1BA	7,105,036	1437	2.0E-04	13,462,393	20	2722.8	0.000000
GRN	-	0		20,567,429	0		n/a
H19	5,074,393	21	4.1E-06	15,493,036	39	64.1	0.000207
HADHA	7,609,500	10	1.3E-06	12,957,929	0	17.0	0.000000
HBA1	15,519,179	2360	1.5E-04	5,048,250	294	767.7	0.000000
HBA2	15,519,179	2360	1.5E-04	5,048,250	294	767.7	0.000000
HBB	15,519,179	963	6.2E-05	5,048,250	60	313.3	0.000000
HBD	11,937,786	175	1.5E-05	8,629,643	5	126.5	0.000000
HBG1	6,863,393	12	1.7E-06	13,704,036	0	24.0	0.000000
HBG2	6,863,393	12	1.7E-06	13,704,036	0	24.0	0.000000
HEXA	11,937,786	1067	8.9E-05	8,629,643	52	771.3	0.000000
HEXB	6,863,393	4	5.8E-07	13,704,036	8	8.0	0.139585
HFE	15,965,893	3376	2.1E-04	4,601,536	0	973.1	0.000000
HIC1	1,550,750	2	1.3E-06	19,016,679	0	24.5	0.000000
HIPK3	-	0		20,567,429	0		n/a
HIRA	14,472,893	189	1.3E-05	6,094,536	0	79.6	0.000000
HLA-A	7,105,036	5220	7.3E-04	13,462,393	30	9890.7	0.000000
HLA-B	13,968,429	5349	3.8E-04	6,599,000	20	2527.0	0.000000
HLA-C	7,105,036	4980	7.0E-04	13,462,393	5	9435.9	0.000000
HLA-DPB1	7,105,036	4920	6.9E-04	13,462,393	0	9322.3	0.000000
HLA-DQA1	5,074,393	300	5.9E-05	15,493,036	260	916.0	0.000000
HLA-DQB1	7,105,036	5200	7.3E-04	13,462,393	10	9852.8	0.000000
HLA-DRB1	7,105,036	5020	7.1E-04	13,462,393	270	9511.7	0.000000
HLA-DRB3	7,105,036	4915	6.9E-04	13,462,393	2	9312.8	0.000000
HLA-DRB4	2,030,643	4900	2.4E-03	18,536,786	0	44729.8	0.000000
HLA-DRB5	2,030,643	4900	2.4E-03	18,536,786	0	44729.8	0.000000
HMBS	-	0		20,567,429	0		n/a
HMGCL	-	0		20,567,429	0		n/a

HMHA1	5,074,393	5	9.9E-07	15,493,036	0	15.3	0.000000
HNF1A	4,028,107	8	2.0E-06	16,539,322	3	32.8	0.000000
HNF4A	-	0		20,567,429	1		n/a
HPRT1	-	0		20,567,429	0		n/a
HRAS	-	0		20,567,429	0		n/a
HSD17B4	-	0		20,567,429	3		n/a
HTT	19,547,286	817	4.2E-05	1,020,143	126	42.6	0.000000
IDS	1,550,750	1	6.4E-07	19,016,679	2	12.3	0.000355
IDUA	-	0		20,567,429	5		n/a
IGH@	13,014,572	1412	1.1E-04	7,552,857	6	819.4	0.000000
IGH@/BCL2	3,581,393	91	2.5E-05	16,986,036	2	431.6	0.000000
IGH@/CCND1	3,581,393	28	7.8E-06	16,986,036	0	132.8	0.000000
IGH@/MYC	2,030,643	4	2.0E-06	18,536,786	0	36.5	0.000000
IGHG1	8,894,036	39	4.4E-06	11,673,393	0	51.2	0.000000
IGHR	5,074,393	18	3.5E-06	15,493,036	0	55.0	0.000000
IKBKAP	11,937,786	445	3.7E-05	8,629,643	0	321.7	0.000000
IKBKG	1,550,750	20	1.3E-05	19,016,679	13	245.3	0.000000
IL2RG	6,863,393	2	2.9E-07	13,704,036	0	4.0	0.018437
IL7R	-	0		20,567,429	0		n/a
ITGA2	7,105,036	1437	2.0E-04	13,462,393	20	2722.8	0.000000
ITGA2B	7,105,036	1804	2.5E-04	13,462,393	20	3418.2	0.000000
ITGB3	7,105,036	1814	2.6E-04	13,462,393	20	3437.1	0.000000
ITGB4	4,028,107	11	2.7E-06	16,539,322	0	45.2	0.000000
JAG1	15,519,179	7	4.5E-07	5,048,250	6	2.3	0.019860
JAK2	15,519,179	928	6.0E-05	5,048,250	49	301.9	0.000000
KAL1	3,581,393	9	2.5E-06	16,986,036	0	42.7	0.000000
KCNE1	7,105,036	48	6.8E-06	13,462,393	5	90.9	0.000000
KCNE2	7,105,036	48	6.8E-06	13,462,393	5	90.9	0.000000
KCNH2	7,105,036	48	6.8E-06	13,462,393	5	90.9	0.000000
KCNJ11	-	0		20,567,429	3		n/a
KCNJ2	5,074,393	43	8.5E-06	15,493,036	5	131.3	0.000000
KCNJ6	-	0		20,567,429	0		n/a
KCNQ1	7,105,036	48	6.8E-06	13,462,393	5	90.9	0.000000

KCNQ1OT1	5,074,393	13	2.6E-06	15,493,036	31	39.7	0.025516
KEL	8,655,786	48	5.5E-06	11,911,643	1	66.1	0.000000
KIR3DL1	5,074,393	30	5.9E-06	15,493,036	3	91.6	0.000000
KIT	11,937,786	110	9.2E-06	8,629,643	56	79.5	0.001097
KMS	1,550,750	1	6.4E-07	19,016,679	0	12.3	0.000005
KRAS	-	0		20,567,429	0		n/a
KRIT1	-	0		20,567,429	0		n/a
KRT10	-	0		20,567,429	0		n/a
KRT14	4,028,107	19	4.7E-06	16,539,322	0	78.0	0.000000
KRT5	4,028,107	19	4.7E-06	16,539,322	0	78.0	0.000000
L1CAM	7,105,036	10	1.4E-06	13,462,393	0	18.9	0.000000
LAMA3	4,028,107	11	2.7E-06	16,539,322	0	45.2	0.000000
LAMB3	4,028,107	11	2.7E-06	16,539,322	0	45.2	0.000000
LAMC2	4,028,107	11	2.7E-06	16,539,322	0	45.2	0.000000
LDLR	-	0		20,567,429	0		n/a
LETM1	-	0		20,567,429	0		n/a
LIMK1	3,581,393	32	8.9E-06	16,986,036	0	151.8	0.000000
LLGL1	1,550,750	2	1.3E-06	19,016,679	0	24.5	0.000000
LMNA	11,937,786	54	4.5E-06	8,629,643	14	39.0	0.000002
LRRK2	5,074,393	1	2.0E-07	15,493,036	2	3.1	0.220037
MAPT	7,105,036	7	9.9E-07	13,462,393	0	13.3	0.000002
MAT1A	-	0		20,567,429	0		n/a
MBP	-	0		20,567,429	0		n/a
MC2R	-	0		20,567,429	3		n/a
MC4R	-	0		20,567,429	1		n/a
MCOLN1	6,863,393	56	8.2E-06	13,704,036	0	111.8	0.000000
MECP2	8,894,036	112	1.3E-05	11,673,393	71	147.0	0.000000
MEFV	10,444,786	183	1.8E-05	10,122,643	100	177.4	0.000000
MEN1	12,683,893	73	5.8E-06	7,883,536	10	45.4	0.000000
MET	8,414,143	4	4.8E-07	12,153,286	1	5.8	0.017889
MFAP4	1,550,750	2	1.3E-06	19,016,679	0	24.5	0.000000
MFN2	8,894,036	44	4.9E-06	11,673,393	6	57.7	0.000000
MGMT	-	0		20,567,429	0		n/a

MKRN3	1,550,750	2	1.3E-06	19,016,679	0	24.5	0.000000
MLH1	19,547,286	376	1.9E-05	1,020,143	99	19.6	0.000000
MLL	8,414,143	26	3.1E-06	12,153,286	9	37.6	0.000000
MLL/AFF1	4,028,107	120	3.0E-05	16,539,322	0	492.7	0.000000
MNX1	-	0		20,567,429	0		n/a
MPZ	8,894,036	17	1.9E-06	11,673,393	3	22.3	0.000000
MSH2	19,547,286	375	1.9E-05	1,020,143	99	19.6	0.000000
MSH6	15,519,179	220	1.4E-05	5,048,250	54	71.6	0.005133
MSI#	13,488,536	359	2.7E-05	7,078,893	9	188.4	0.000000
MSRA	-	0		20,567,429	0		n/a
MT-ATP6	10,653,250	381	3.6E-05	9,914,179	55	354.6	0.000000
MT-CO1	-	0		20,567,429	1		n/a
MT-CO2	-	0		20,567,429	1		n/a
MT-CO3	-	0		20,567,429	1		n/a
MT-CYB	4,028,107	24	6.0E-06	16,539,322	0	98.5	0.000000
MT-deletion#	9,102,500	119	1.3E-05	11,464,929	2	149.9	0.000000
MTHFR	9,102,500	293	3.2E-05	11,464,929	0	368.4	0.000000
MTM1	-	0		20,567,429	3		n/a
MT-ND1	10,653,250	66	6.2E-06	9,914,179	10	61.4	0.000000
MT-ND4	10,653,250	275	2.6E-05	9,914,179	10	255.9	0.000000
MT-ND5	13,488,536	30	2.2E-06	7,078,893	4	15.7	0.000372
MT-ND6	6,625,143	42	6.3E-06	13,942,286	10	88.4	0.000000
MT-RNR1	3,581,393	3	8.4E-07	16,986,036	1	14.2	0.000009
MT-TK	17,516,643	457	2.6E-05	3,050,786	62	79.6	0.006159
MT-TL1	10,653,250	331	3.1E-05	9,914,179	5	308.0	0.000000
MUTYH	13,488,536	277	2.1E-05	7,078,893	27	145.4	0.000000
MYBPC3	5,074,393	4	7.9E-07	15,493,036	0	12.2	0.000005
MYC	10,444,786	37	3.5E-06	10,122,643	0	35.9	0.000000
MYCN	9,102,500	45	4.9E-06	11,464,929	0	56.7	0.000000
MYH7	5,074,393	3	5.9E-07	15,493,036	0	9.2	0.000105
MYOT	-	0		20,567,429	0		n/a
NAGLU	-	0		20,567,429	1		n/a
NDN	1,550,750	2	1.3E-06	19,016,679	0	24.5	0.000000

NDP	4,028,107	6	1.5E-06	16,539,322	0	24.6	0.000000
NF1	2,030,643	13	6.4E-06	18,536,786	0	118.7	0.000000
NF2	7,105,036	21	3.0E-06	13,462,393	11	39.8	0.000000
NIPA1	6,863,393	1	1.5E-07	13,704,036	1	2.0	0.271119
NOD2	5,074,393	50	9.9E-06	15,493,036	8	152.7	0.000000
NOG	-	0		20,567,429	0		n/a
NOTCH3	6,058,750	35	5.8E-06	14,508,679	70	83.8	0.014251
NPC1	8,414,143	57	6.8E-06	12,153,286	3	82.3	0.000000
NR0B1	-	0		20,567,429	0		n/a
NR4A2	-	0		20,567,429	0		n/a
NSD1	3,581,393	8	2.2E-06	16,986,036	0	37.9	0.000000
OCRL	-	0		20,567,429	0		n/a
OTC	17,516,643	24	1.4E-06	3,050,786	6	4.2	0.113336
OTOF	-	0		20,567,429	0		n/a
PABPN1	4,028,107	21	5.2E-06	16,539,322	0	86.2	0.000000
PAFAH1B1	10,444,786	12	1.1E-06	10,122,643	0	11.6	0.000009
PAH	4,028,107	1	2.5E-07	16,539,322	0	4.1	0.016474
PALB2	-	0		20,567,429	0		n/a
PANK2	-	0		20,567,429	0		n/a
PARK2	5,074,393	5	9.9E-07	15,493,036	4	15.3	0.000531
PARK7	-	0		20,567,429	1		n/a
PAX6	1,550,750	6	3.9E-06	19,016,679	0	73.6	0.000000
PAX7/FOXO1A	5,074,393	3	5.9E-07	15,493,036	0	9.2	0.000105
PDCD6	-	0		20,567,429	0		n/a
PDHA1	5,074,393	1	2.0E-07	15,493,036	0	3.1	0.047209
PEX1	1,550,750	5	3.2E-06	19,016,679	3	61.3	0.000000
PEX7	-	0		20,567,429	3		n/a
PINK1	-	0		20,567,429	2		n/a
PITX2	-	0		20,567,429	0		n/a
PKP2	5,074,393	2	3.9E-07	15,493,036	0	6.1	0.002229
PLA2G6	-	0		20,567,429	0		n/a
PML/RARA	15,519,179	111	7.2E-06	5,048,250	0	36.1	0.000000
PMM2	-	0		20,567,429	8		n/a

PMP22	15,519,179	257	1.7E-05	5,048,250	43	83.6	0.000000
PMS2	8,655,786	13	1.5E-06	11,911,643	19	17.9	0.088099
POLG	5,074,393	3	5.9E-07	15,493,036	3	9.2	0.013475
POU1F1	-	0		20,567,429	0		n/a
PPOX	-	0		20,567,429	0		n/a
PPP2R2B	-	0		20,567,429	0		n/a
PPT1	-	0		20,567,429	3		n/a
PRF1	-	0		20,567,429	0		n/a
PRNP	7,105,036	24	3.4E-06	13,462,393	31	45.5	0.005327
PROP1	-	0		20,567,429	0		n/a
PRPSAP2	1,550,750	2	1.3E-06	19,016,679	0	24.5	0.000000
PRSS1	2,030,643	4	2.0E-06	18,536,786	0	36.5	0.000000
PSEN1	5,074,393	30	5.9E-06	15,493,036	10	91.6	0.000000
PSEN2	-	0		20,567,429	0		n/a
PTEN	10,444,786	21	2.0E-06	10,122,643	2	20.4	0.000000
PTPN11	2,030,643	17	8.4E-06	18,536,786	0	155.2	0.000000
PYGM	2,030,643	1	4.9E-07	18,536,786	1	9.1	0.000991
RAF1	-	0		20,567,429	0		n/a
RAG1	-	0		20,567,429	0		n/a
RAG2	-	0		20,567,429	0		n/a
RAI1	3,581,393	33	9.2E-06	16,986,036	0	156.5	0.000000
RARA	1,550,750	9	5.8E-06	19,016,679	0	110.4	0.000000
RB1	8,655,786	147	1.7E-05	11,911,643	31	202.3	0.000000
RET	15,519,179	119	7.7E-06	5,048,250	17	38.7	0.000043
RHCE	6,625,143	43	6.5E-06	13,942,286	1	90.5	0.000000
RHD	13,488,536	63	4.7E-06	7,078,893	7	33.1	0.000000
RUNX1/RUNX1T1	8,655,786	30	3.5E-06	11,911,643	0	41.3	0.000000
RYR1	2,030,643	4	2.0E-06	18,536,786	13	36.5	0.000005
SAMD12	-	0		20,567,429	0		n/a
SBDS	2,030,643	2	9.8E-07	18,536,786	0	18.3	0.000000
SCN1A	5,074,393	6	1.2E-06	15,493,036	3	18.3	0.000011
SCN4A	6,058,750	9	1.5E-06	14,508,679	1	21.6	0.000000
SCN5A	5,074,393	43	8.5E-06	15,493,036	5	131.3	0.000000

SCNN1D	1,550,750	2	1.3E-06	19,016,679	0	24.5	0.000000
SDHB	6,863,393	37	5.4E-06	13,704,036	59	73.9	0.010366
SDHD	6,863,393	19	2.8E-06	13,704,036	37	37.9	0.064679
SEMA5A	-	0		20,567,429	0		n/a
SEPN1	5,074,393	2	3.9E-07	15,493,036	0	6.1	0.002229
SERPINA1	11,133,143	1288	1.2E-04	9,434,286	0	1091.5	0.000000
SERPING1	-	0		20,567,429	30		n/a
SFTPB	6,863,393	4	5.8E-07	13,704,036	0	8.0	0.000340
SGSH	-	0		20,567,429	8		n/a
SH2D1A	2,030,643	2	9.8E-07	18,536,786	0	18.3	0.000000
SHOX	1,550,750	18	1.2E-05	19,016,679	0	220.7	0.000000
SKI	1,550,750	2	1.3E-06	19,016,679	0	24.5	0.000000
SLBP	-	0		20,567,429	0		n/a
SLC14A1	8,655,786	48	5.5E-06	11,911,643	1	66.1	0.000000
SLC26A4	2,030,643	41	2.0E-05	18,536,786	0	374.3	0.000000
SMAD4	6,863,393	1	1.5E-07	13,704,036	1	2.0	0.271119
SMARCB1	5,074,393	2	3.9E-07	15,493,036	0	6.1	0.002229
SMCR	6,863,393	1	1.5E-07	13,704,036	0	2.0	0.135785
SMN1	15,519,179	559	3.6E-05	5,048,250	138	181.8	0.000106
SMPD1	5,074,393	79	1.6E-05	15,493,036	0	241.2	0.000000
SNAP29	1,550,750	2	1.3E-06	19,016,679	0	24.5	0.000000
SNCA	6,863,393	2	2.9E-07	13,704,036	5	4.0	0.156034
SNCAIP	-	0		20,567,429	0		n/a
SNRPN	19,547,286	460	2.4E-05	1,020,143	8	24.0	0.000103
SOD1	13,968,429	19	1.4E-06	6,599,000	15	9.0	0.019122
Somatichypermutation#	1,550,750	1	6.4E-07	19,016,679	0	12.3	0.000005
SOS1	-	0		20,567,429	0		n/a
SPAST	8,894,036	12	1.3E-06	11,673,393	1	15.7	0.000002
SPG3A	6,863,393	1	1.5E-07	13,704,036	1	2.0	0.271119
SPON2	-	0		20,567,429	0		n/a
SPTLC1	6,863,393	13	1.9E-06	13,704,036	6	26.0	0.000002
SRD5A2	-	0		20,567,429	0		n/a
SRY	14,472,893	360	2.5E-05	6,094,536	6	151.6	0.000000

SS18	1,550,750	2	1.3E-06	19,016,679	0	24.5	0.000000
STK11	6,863,393	29	4.2E-06	13,704,036	13	57.9	0.000000
STR#	17,516,643	117	6.7E-06	3,050,786	65	20.4	0.000000
STS	1,550,750	5	3.2E-06	19,016,679	0	61.3	0.000000
STX1A	1,550,750	2	1.3E-06	19,016,679	0	24.5	0.000000
Subteldeletion#	15,519,179	1450	9.3E-05	5,048,250	0	471.7	0.000000
SURF1	5,074,393	6	1.2E-06	15,493,036	2	18.3	0.000002
TACC3	-	0		20,567,429	0		n/a
TBP	6,863,393	5	7.3E-07	13,704,036	0	10.0	0.000046
TCF3	1,550,750	8	5.2E-06	19,016,679	9	98.1	0.000000
TCF3/PBX1	4,028,107	120	3.0E-05	16,539,322	0	492.7	0.000000
TERT	-	0		20,567,429	0		n/a
TGFBR1	6,863,393	2	2.9E-07	13,704,036	3	4.0	0.195690
TGFBR2	6,863,393	2	2.9E-07	13,704,036	3	4.0	0.195690
TH	-	0		20,567,429	0		n/a
TNFRSF13B	1,550,750	2	1.3E-06	19,016,679	0	24.5	0.000000
TNFRSF18	1,550,750	2	1.3E-06	19,016,679	0	24.5	0.000000
TNFRSF1A	6,863,393	58	8.5E-06	13,704,036	44	115.8	0.000000
TNFRSF4	1,550,750	2	1.3E-06	19,016,679	0	24.5	0.000000
TOR1A	3,581,393	14	3.9E-06	16,986,036	20	66.4	0.000000
TP53	15,519,179	63	4.1E-06	5,048,250	12	20.5	0.014417
TP73	1,550,750	2	1.3E-06	19,016,679	0	24.5	0.000000
TPM3	2,030,643	1	4.9E-07	18,536,786	2	9.1	0.004522
TPMT	3,581,393	120	3.4E-05	16,986,036	1	569.1	0.000000
TPP1	1,550,750	8	5.2E-06	19,016,679	11	98.1	0.000000
TRAPPC2	-	0		20,567,429	2		n/a
TRB@	9,102,500	1164	1.3E-04	11,464,929	0	1466.1	0.000000
TRG@	12,683,893	1448	1.1E-04	7,883,536	3	900.0	0.000000
TRPS1	1,550,750	5	3.2E-06	19,016,679	0	61.3	0.000000
TRPV1	1,550,750	2	1.3E-06	19,016,679	0	24.5	0.000000
TSC1	8,894,036	10	1.1E-06	11,673,393	3	13.1	0.000752
TSC2	13,968,429	14	1.0E-06	6,599,000	3	6.6	0.064690
TTN	-	0		20,567,429	0		n/a

TTR	11,937,786	5	4.2E-07	8,629,643	0	3.6	0.026932
TWIST1	8,414,143	57	6.8E-06	12,153,286	56	82.3	0.000461
TWISTNB	1,550,750	2	1.3E-06	19,016,679	0	24.5	0.000000
TYR	2,030,643	1	4.9E-07	18,536,786	0	9.1	0.000109
UBE3A	19,547,286	360	1.8E-05	1,020,143	5	18.8	0.000135
UCHL1	-	0		20,567,429	0		n/a
UGT1A1	7,105,036	171	2.4E-05	13,462,393	3	324.0	0.000000
UNC13D	2,030,643	3	1.5E-06	18,536,786	0	27.4	0.000000
VHL	10,444,786	35	3.4E-06	10,122,643	63	33.9	0.000002
VKORC1	5,074,393	60	1.2E-05	15,493,036	35	183.2	0.000000
VWF	1,550,750	5	3.2E-06	19,016,679	0	61.3	0.000000
WAS	2,030,643	3	1.5E-06	18,536,786	0	27.4	0.000000
WFS1	6,863,393	4	5.8E-07	13,704,036	2	8.0	0.010842
WHSC1	1,550,750	3	1.9E-06	19,016,679	0	36.8	0.000000
WHSC2	-	0		20,567,429	0		n/a
WT1	5,074,393	2	3.9E-07	15,493,036	11	6.1	0.024575
YWHAE	3,581,393	6	1.7E-06	16,986,036	0	28.5	0.000000