Deliver specimens to:
Central Specimen Reception (CSR) Sydpath
St Vincent's Hospital, Xavier Building
Level 6, 390 Victoria St
Darlinghurst NSW 2010 Australia
P +61 2 8382 9100

This form is available to complete online at
my.genome.one/test-request

Genome.One Pty Ltd
ACN 608 029 732
370 Victoria St
Darlinghurst NSW 2010
Sydney Australia
P +61 2 9359 8002
F +61 2 9359 8033
E enquiries@genome.one

Genome.One Test Request Form

This document contains:

- Information about our incidental findings and secondary analysis policy on the next page.
- Three forms within designed for a trio request. The first form is used for the proband, and the following two forms are used for additional family members if appropriate.
- If you are only making a request for a single patient, please complete the first form and discard the following two forms for family members.
- You can also find information about our privacy collection statement and consent on the final page of this document.

Incidental Findings Policy

Genome.One uses an OPT-IN approach to determine patient consent for incidental findings.

An incidental finding is defined by Genome.One as:

- A variant in a gene NOT relevant to the phenotype of the patient;

AND

- A Class 4 or 5 variant in ClinVar in a gene described as medically actionable; the ACMG59 (Kalia et al.; 2017) where review by Genome.One is in agreement with their likely pathogenic classifications, and where variant zygosity fits the known inheritance pattern for the disorder.

If a patient has NOT opted in, then NO incidental findings will be reported, and the report will explicitly state that incidental findings (including ACMG59) have not been actively examined.

In the rare event incidental findings outside the ACMG59 (or any incidental findings in a patient under the age of 16 years) are identified they will be managed on a case-by-case discussion between the laboratory and referring clinician, with the option of a multidisciplinary team meeting.

Genome.One can also offer a secondary analysis

A secondary analysis (of additional genes not relevant to the phenotype or family history) can be requested in individuals for the ACMG59 (16 years and over) and/or pharmacogenomics (independent of age).

USE THIS PAGE FOR THE PROBAND

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Patient details

First Name | Sex

Last Name | Date of Birth DD/MMM/YY

Ethnicity (if known)

My Health Record

☐ Do not send results report to My Health Record

The patient's Medicare or Department of Veteran Affairs details are required to send reports to My Health Record

Medicare card or DVA Health card number (include expiry)

Test requested

Primary analysis

Whole genome sequencing
☐ Analyse all genes
☐ Analyse a panel of genes

Whole exome sequencing
☐ Analyse all genes
☐ Analyse a panel of genes

Write in name of Genome.One panel requested, OR, a list of genes you want analysed (or email to enquiries@genome.one).

☐ If no likely pathogenic or pathogenic variant (Class 4/5) are identified in the panel, would you like a reflex analysis for all genes? (Additional costs will apply)

Secondary analysis (additional costs may apply)

☐ Pharmacogenomics (only if whole genome sequencing is selected)
☐ ACMG59

Additional information

Consent

Genomic consent

Either
☐ I confirm that the patient/parent/guardian has provided informed consent for genomic testing; *OR*
☐ I would like a genetic counsellor from Genome.One to obtain consent (a genetic counsellor from Genome.One will then call you to discuss this.) **You do not have to complete the remaining consent section**

Incidental findings

I confirm that the patient is 16 years or over and has *OPTED IN* to receive incidental findings limited to **ACMG59** Class 4 or 5 variants not relevant to the primary analysis (i.e. they wish to receive this information).
I confirm that the patient understands that in the rare event incidental findings *outside* the **ACMG59** are identified (or any incidental findings in a patient under the age of 16 years), they will be managed on a case-by-case discussion between the laboratory and referring clinician, with the option of a multidisciplinary team meeting.
☐ Yes ☐ No

Secondary analysis

I confirm that this patient has *OPTED IN* and provided consent to have additional analysis for **ACMG59** (available only if patient over 16 years of age).
☐ Yes ☐ No

I confirm that this patient has requested and provided consent to have additional analysis for **Pharmacogenomics** (independent of age)
☐ Yes ☐ No

Disclosure of information to relatives

I confirm that the patient/parent/guardian understands that their and/or their child's test results may have implications for blood relatives (e.g. parents, siblings, children). If the results have implications for their blood relatives, the patient/parent/guardian consents to their and/or their child's results being given to their blood relatives and the health professionals involved in their care.
☐ Yes ☐ No

Research and future use of genetic information

I confirm that the patient/parent/guardian has provided consent for their and/or their child's deidentified genetic information to be placed in reference databases and shared with researchers and clinicians for the purpose of research into the genetic causes of disease, to improve genomic pathology services and for the development of drugs and therapies.
☐ Yes ☐ No

Privacy

☐ I confirm that the patient/parent/guardian has been provided with a copy of Genome.One's Privacy Policy (www.genome.one/privacy-policy) and Privacy Collection Statement.

USE THIS PAGE FOR THE PROBAND

Clinical information

Provide clinical notes and the diagnosis.
Comprehensive notes increase the chance of a successful diagnosis.

Family history.
Please draw a pedigree and/or describe the family history

Provide results from relevant investigations.
For example: genetic tests, other pathology imaging.

☐ Is consanguinity present?

Family members tested as part of this analysis

Separate samples will follow for these additional family members. Separate request forms are required for each family member.

Name	Name
Relationship	Relationship
Date of Birth DD/MMM/YY	Date of Birth DD/MMM/YY

Requesting clinician

Provider number	Medical specialty
First Name	Name of organisation
Last Name	Phone Number
Email	Fax (optional)

Billing

Select one:
☐ Institution
☐ Private – I confirm that the person below will pay for the test:
☐ Other:

First Name	Last Name
Email	Mobile Number *(The mobile number entered will be used as the password to open the invoice sent to the payer)*

Copy report to

An encrypted copy of the report will be sent by email.

First Name	Last Name
Name of organisation	Email

Collector declaration and sample details

I certify that I collected the accompanying specimens from the above, whose identity was confirmed by enquiry and/or examination of their name band and that I labelled the specimens immediately following collection.

Collector's name	Collection date DD/MMM/YY
Collection time	Signature

Sample type/size ☐ EDTA Blood: 2 mL (paediatric), 5 mL (adults) ☐ DNA: Conc 10-100 ng/uL, Vol 100 uL

Stop here if this request is only for a singleton patient; if testing is on a family study e.g. trio, please continue

USE THIS PAGE FOR AN ADDITIONAL FAMILY MEMBER BEING TESTED

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Family member details

First Name | Sex

Last Name | Date of Birth DD/MMM/YY

Ethnicity (if known)

Relation to proband

My Health Record
☐ Do not send results report to My Health Record

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Medicare card or DVA Health card number (include expiry)

Test requested

Primary analysis

Whole genome sequencing
☐ Analyse all genes
☐ Analyse a panel of genes to match the proband

Whole exome sequencing
☐ Analyse all genes
☐ Analyse a panel of genes to match the proband

Secondary analysis (additional costs may apply)
☐ Pharmacogenomics (only if whole genome sequencing is selected)
☐ ACMG59

Additional information

Consent

Genomic consent:
Either
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Incidental findings
I confirm that the family member is 16 years or over and has *OPTED IN* to receive incidental findings limited to **ACMG59** Class 4 or 5 variants not relevant to the primary analysis (i.e. they wish to receive this information).
I confirm that the family member understands that in the rare event incidental findings *outside* the **ACMG59** are identified (or any incidental findings in a patient under the age of 16 years), they will be managed on a case-by-case discussion between the laboratory and referring clinician, with the option of a multidisciplinary team meeting.
☐ Yes ☐ No

Secondary analysis
I confirm that this family member has *OPTED IN* and provided consent to have additional analysis for **ACMG59** (available only if the family member is over 16 years of age).
☐ Yes ☐ No

I confirm that this family member has requested and provided consent to have additional analysis for **Pharmacogenomics** (independent of age)
☐ Yes ☐ No

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I confirm that the family member's test results may have implications for blood relatives (e.g. parents, siblings, children). If the results have implications for their blood relatives, the family member consents to their results being given to their blood relatives and the heath professionals involved in their care.
☐ Yes ☐ No

Research and future use of genetic information
I confirm that the family member has provided consent for their deidentified genetic information to be placed in reference databases and shared with researchers and clinicians for the purpose of research into the genetic causes of disease, to improve genomic pathology services and for the development of drugs and therapies.
☐ Yes ☐ No

Privacy
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Clinical information

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Requesting clinician

Provider number	Medical specialty
First Name	Last Name
Name of organisation	Phone Number
Email	Fax (optional)

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Provider number	Medical specialty
First Name	Last Name
Name of organisation	Phone Number
Email	Fax (optional)

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Privacy Collection Statement and Consent

- Your requesting clinician has requested a pathology report from a Genome.One genetic pathologist. You will be asked to provide a blood sample, and may also be asked to provide a saliva or a cheek brush sample. The pathology report will be based on the results of a screening test of genetic information derived from the sample collected from you. Please review the Patient Information Sheet for more information about the screening test and the pathology report from Genome.One.
- We collect personal information about you when we receive a request for a genomic pathology report for diagnostic or health management purposes. We will usually collect your personal information (including information about your health) from your requesting clinician or directly from you.
- We collect personal information about you to provide you with genomic pathology services. If we do not collect personal information about you, we may be unable to provide you with the services your requesting clinician has requested.
- When we receive a request for a genomic pathology report, we collect personal information such as your name, date of birth, address and other contact details. We collect information about you from the request form including your medical history; results of previous tests and investigations; your family medical history; and details about your treating doctors and other health professionals involved in your care.
- We will disclose your personal information, typically in the form of a genomic pathology report, to your requesting clinician. This report may also come with additional information relevant to your health.
- Genome.One works with reputable contractors and service providers. Those organisations provide us with genomic medicine, scientific and technical services necessary to provide our services. We require our service providers to comply with Genome.One's Privacy Policy and Australian privacy laws (or the foreign equivalent, if they are located overseas).
- Genome.One does not typically or routinely disclose personal information to overseas recipients, unless your clinician is located overseas. However, genetic data derived from the blood sample you provide is held on servers located in the United States of America for storage and for highly-specialised data processing and analysis. Before that data is released, information that identifies you is removed and replaced with a secure code. Information which is derived from the blood sample you provide may be collected in databanks managed by Genome.One or external organisations. These databanks are used to improve genomic pathology services to other individuals; to further clinical research; and may be used by researchers and industry to develop new drugs, treatments and therapies.
- Genome.One handles your personal information in accordance with its Privacy Policy. Please ask your requesting clinician to provide you with a copy of our Privacy Policy or obtain a copy at http://genome.one. The Privacy Policy includes more detailed information about the collection, use, disclosure and security of your personal information. The Privacy Policy includes information about how you may request access to, and correction of, personal information we hold about you. The Privacy Policy also includes information about how you may make a complaint about how your personal information has been handled.

You can access the full Genome.One privacy policy on our website www.genome.one