Part A: Genomic Testing Request Form

PATIENT DETAILS	
MRN:	Phone/ Mobile:
Surname:	Address:
Given Name: DOB:	
Gender: ☐ Female ☐ Male ☐ Unknown	Email:

REQUESTING DOCTOR	
Name:	Provider Number:
Address:	☐ Email to:
Phone/ Mobile:	☐ Hard copy:
Signature:	

COPY REPORT TO	
Doctor:	☐ Email copy to:
Phone/ Mobile:	☐ Hard copy to:

TEST REQUESTED

☐ **Whole Exome** analysis

☐ **Gene panel only** analysis (tick box/es on page 2- clinical indications or attach a gene list)

☐ **Gene panel analysis,** with whole exome analysis *if* nothing clinically relevant found in the panel

☐ **Re-analysis of Whole Exome**, please specify reason for re-analysis under Clinical Information

☐ **Proband only**

☐ **Family**

Indicate ☐Proband ☐ Mother ☐ Father ☐ Other

If not the proband, please include the proband's Full Name: DOB:

SPECIMEN INFORMATION (Collector / Sender to complete)			
Print Name: Signature:		Date and time of collection:	
EDTA Whole Blood *(5-10mls for adults, 2-5mls for children)*	Number of tubes collected:		
Extracted DNA *(50-100ng/µl, total volume ≥50µl)*	Concentration:	Elution Buffer:	Total Volume:
Other sample types *(i.e. buccal swab, saliva)*, details:			

For ACT Pathology collection centres:

- Collect 1 x 5-10ml EDTA (adults) or 1x 2-5ml EDTA (children)
- Register test as "CCG Test". Refer to Kestral ALT-9 for more information

For all other collection centres:

- Collect 1 x 5-10ml EDTA (adults) or 1x 2-5ml EDTA (children)
- Send samples to: Canberra Clinical Genomics, The Australian National University, Hugh Ennor Building, 117 Garran Rd, ACTON, ACT, 2601.

For any issues and/ or enquires please contact us on (02) 5124 5630

CLINICAL INDICATIONS (Please tick relevant box/es)

Developmental / Congenital
- ☐ Developmental Delay / Intellectual Disability
- ☐ Dysmorphism/s
- ☐ Floppy Infant
- ☐ IUGR and IGF abnormalities
- ☐ RASopathies
- ☐ Paediatric Disorder – Specific or Syndromic
- ☐ Other (specify next page)

Neurological
- ☐ Ataxia / Movement / Tone Disorder
- ☐ Hereditary Spastic Paraplegia
- ☐ Autism
- ☐ Brain Malformation
- ☐ Inherited White Matter Disorder
- ☐ Epilepsy
- ☐ Dysautonomia
- ☐ Pain Syndrome
- ☐ Hereditary Neuropathy of PNS
- ☐ Familial Dementia
- ☐ Degenerative Brain Disorder
- ☐ Parkinson Disease
- ☐ Retinal Disorder
- ☐ Eye Disorder, other
- ☐ Deafness
- ☐ Motor Neuron Disease
- ☐ Other (specify next page)

Musculoskeletal
- ☐ Craniofacial Abnormalities
- ☐ Connective Tissue Disorder
- ☐ Muscular Dystrophy
- ☐ Rhabdomyolysis and Metabolic Muscle Disorders
- ☐ Skeletal Disorder
- ☐ Arthrogryposis
- ☐ Other (specify next page)

Immunological
- ☐ Inflammatory / Autoimmune Disorder
- ☐ Primary Immune Deficiency
- ☐ Other (specify below)

Coagulation/Blood
- ☐ Bleeding disorder
- ☐ Thrombotic disorder
- ☐ Haemoglobinopathy (Thalassaemia, Haemoglobin Variant)
- ☐ Anaemia / Red Cell Disorder
- ☐ Other (specify next page)

Endocrine
- ☐ Hypothalamic / Pituitary
- ☐ Calcium Homeostasis Disorder
- ☐ Diabetes
- ☐ Severe early-onset obesity
- ☐ Other (specify next page)

Cardiovascular
- ☐ Cardiomyopathy
- ☐ Cardiac Arrhythmia / SCD
- ☐ Dyslipidaemia
- ☐ Vascular Abnormalities / Primary Lymphoedema
- ☐ Congenital Heart Defect
- ☐ Hypertension (Left sided / Pulmonary)
- ☐ Other (specify next page)

Respiratory
- ☐ Cystic Fibrosis
- ☐ COPD / Non-CF bronchiectasis
- ☐ Restrictive Lung Disease
- ☐ Ciliary Dyskinesia / Laterality Disorder
- ☐ Surfactant Deficiency
- ☐ Other (specify next page)

Renal
- ☐ Cystic Kidney Disease
- ☐ Haematuria / Proteinuria
- ☐ Glomerular Disease
- ☐ Tubulointerstitial Kidney Disease
- ☐ Renal Tubulopathies
- ☐ Nephrocalcinosis or Nephrolithiasis
- ☐ Renal Ciliopathies / Renal and Urinary tract malformations
- ☐ Unexplained End Stage Renal Disease
- ☐ Other (specify next page)

Other Organs
- ☐ Polycystic Liver Disease
- ☐ Liver disorder, other
- ☐ Pancreatic disorder / Pancreatitis
- ☐ Other (specify next page)

Metabolic
- ☐ Inborn Error of Metabolism / Mitochondrial Disorder
- ☐ Lysosomal Storage Disorder
- ☐ Peroxisomal Disorder
- ☐ Iron Metabolism Disorder
- ☐ Other (specify next page)

Gastrointestinal
- ☐ Dysmotility
- ☐ Epithelial Barrier Disorder / Diarrhoeal disorder
- ☐ GIT malformation/s
- ☐ Other (specify next page)

Dermatological
- ☐ Epidermolysis Bullosa
- ☐ Autoimmune Skin Disorder
- ☐ Palmoplantar Keratodermas
- ☐ Pigmentary Skin Disorder
- ☐ Vascular Skin Disorder
- ☐ Other (specify next page)

Cancer Susceptibility
- ☐ Breast & Ovarian Cancer
- ☐ Bowel Cancer / Lynch syndrome
- ☐ Renal Cancer
- ☐ Head & Neck
- ☐ Multiple Endocrine Tumour
- ☐ Melanoma
- ☐ Multiple Tissues
- ☐ Other (specify next page)

Sexual Developmental
- ☐ Primary Ovarian Insufficiency
- ☐ Other (specify below)

Sudden Death
- ☐ Sudden Infant Death (SIDS)
- ☐ Sudden Unexplained Death

For a specific gene panel please attach the gene list to the request form

DETAILED CLINICAL HISTORY / DIFFERENTIAL DIAGNOSIS

See over page for helpful hints

PREVIOUS GENETIC TESTING AND/ OR CLINICALLY RELEVANT RESULTS

Please include the test, laboratory and result

FAMILY HISTORY (Draw pedigree below or attach a copy)

See over page for helpful hints

Are family members available for testing: Mother ☐ Yes ☐ No Father ☐ Yes ☐ No Other ☐:

Reason for test: ☐ Diagnostic ☐ Predictive ☐ Family studies

Known Consanguinity: ☐ Yes ☐ No If yes, please describe degree of relation:

REQUESTING HEALTH PROFESSIONAL

Full Name:	Position/Department/Institution:
Signature:	Date:

HELPFUL HINTS

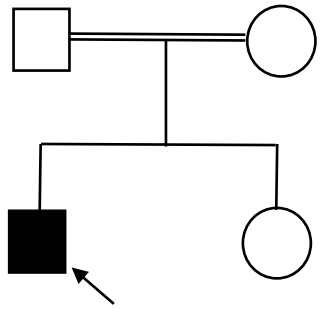
Clinical Description

- A *detailed* clinical description can significantly improve the chance of finding a genetic diagnosis
- *Rare* or unusual signs or symptoms can be most helpful for genotype:phenotype correlation
- Please add *extra clinical notes* to the request form if available
- Human Phenotype Ontology (HPO) terms provide a standardized, hierarchical vocabulary of phenotypic abnormalities encountered in human disease. They can be found at this website: https://hpo.jax.org/app/
- A Clinical Geneticist can help with this

Family History

- Genetics is a science that *involves families*
- Clinical Genomics includes filtering through ≈25,000 DNA variations per patient. It is a 'needle in the haystack' problem. Three things help genome scientists find an answer:
    1. Detailed *clinical description* (see points above)
    2. Clinically annotated *family pedigree* (see 2 examples below), and
    3. *Inclusion of relatives* in the testing process. A *distant relative* with the same condition can be most valuable for the variant filtering process

Example 1. Unaffected (consanguineous) parents, 1 affected male offspring, 1 unaffected female offspring

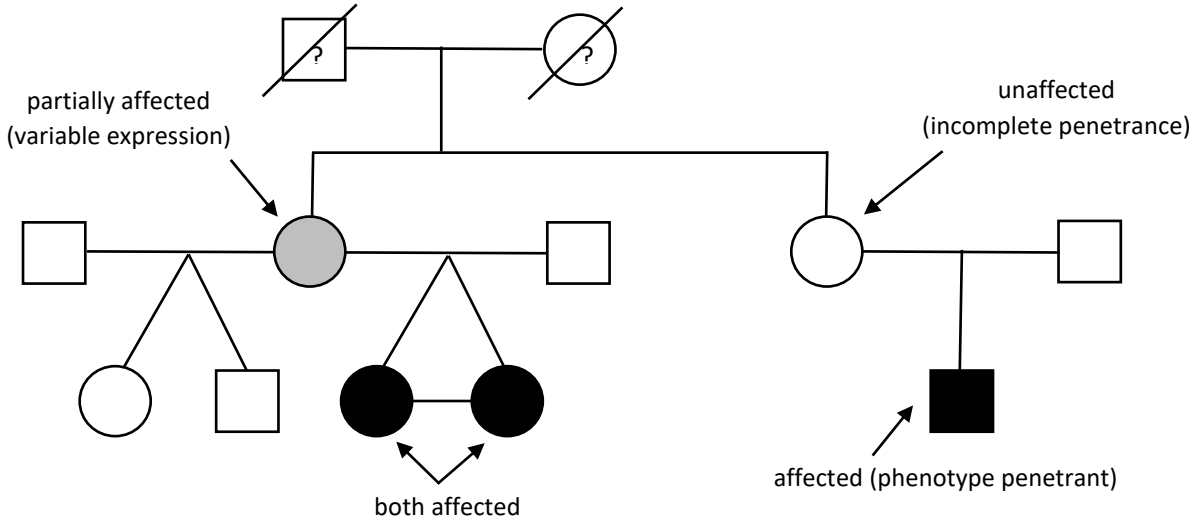


Male Proband (affected)

Possible modes of inheritance

- Autosomal Recessive with both parents' carriers *(most likely scenario due to consanguinity)*
- *De Novo* (new) dominant variant in male offspring
- Autosomal Dominant with incomplete penetrance
- X-Linked Recessive inheritance from mother
- Complex inheritance involving more than 1 gene

Example 2. Multigenerational family with two fathers & one mother. Affected monozygotic (identical) twins from one side with a partially affected mother (variable expressivity) and cousin also affected. Unaffected dizygotic (fraternal) twins on the other side. Deceased grandparents with unknown phenotype.



Part B: Genomic Testing Consent Form: ADULT

PATIENT DETAILS	
MRN:	Phone/ Mobile:
Surname:	Address:
Given Name: DOB:	
Gender: ☐ Female ☐ Male ☐ Unknown	Email:

PATIENT CONSENT

I understand:

- My DNA will be tested, by whole exome sequencing (WES), for genes associated with my / my child's condition.
- This test is NOT a general health test and will not identify all gene changes that could contribute to health problems in the future.
- Possible results: A range of clinical results may be reported. The results may include DNA variation that is well understood *or* results that are currently uncertain which may be clarified in the future or require further testing to interpret.
- There is a small chance a genetic variant may be identified that is associated with an unrelated condition that may develop in the future, or that may reveal carrier status of an unrelated condition, these are defined as *incidental findings*. Incidental findings are rare (found in approximately 1% of cases). Only incidental findings that have a >90% confidence of being clinically relevant are reported.
- Test results may have implications for the health care of my blood relatives.
- Testing may reveal non-paternity or non-maternity of a presumed natural parent.
- Testing will not currently affect the ability to obtain health insurance but may affect applications for some types of risk-rated insurances such as life and income protection insurance.
- My DNA sample and genomic data will be stored in accordance with national diagnostic laboratory guidelines.
- My genomic data and associated healthcare information can be used and disclosed in accordance with applicable health privacy laws.
- Testing is voluntary and I can withdraw or cancel testing at any stage.
- My de-identified genomic data and associated health information may be submitted to national or international clinical databases (restricted access).
- The American College of Medical Genetics and Genomics (ACMG) recommends the *additional screening of 59 specific genes* when an exome is clinically analysed. The conditions linked to pathological DNA variants in these genes are well understood and sufficiently serious to recommend clinical follow up and could be used in the future to inform clinical treatment. Only DNA variants with a >90% likelihood of being clinically relevant are reported. This occurs in a small minority of cases.

  Do you want this test to extend to these 59 genes (refer to gene listing by disorder below*) ☐ **Yes** ☐ **No**

- Sharing information with health practitioners involved in the care of the patient and genetic relatives is important in individual and family care. It reduces the work required for informing relevant practitioners and allows access to information that is relevant for other family members.

  ☐ **I consent** ☐ **I do not consent - to share my information with other relevant health practitioners.**

I consent to the genomic testing described above. Genomic testing has been explained to me by a health professional and I have had the opportunity to ask questions and I am satisfied with the explanations.

Patient / Parent / Guardian Name	Patient / Parent/ Guardian Signature	Date
Health Professional Name	Health Professional Signature	Date

***ACMG 59 genes** (grouped by main associated disorder):
***Cardiovascular disease risk genes**: LDLR, APOB, PCSK9, KCNH2, KCNQ1, SCN5A, MYBPC3, MYH11, MYH7, MYL2, MYL3, TNNI3, TNNT2, RYR2, PRKAG2, DSC2, DSG2, DSP, ACTA2, ACTC1, LMNA, PKP2, SMAD3, TMEM43, TPM1*
***Cancer risk genes**: BRCA1, BRCA2, APC, MLH1, MSH2, MSH6, MUTYH, BMPR1A, PMS2 TP53, RET, RB1, VHL, WT1, MEN1, NF2, SDHAF2, SDHB, SDHC, SDHD, SMAD4, STK11, TGFBR1, TGFBR2*
***Musculoskeletal disorder genes**: FBN1, CACNA1S, RYR1, COL3A1*
***Neurological / Intellectual disorder risk genes**: PTEN, TSC1, TSC2*
***Biochemical disorder genes**: ATP7B, GLA, OTC*

Part C: Genomic Testing Consent Form: PAEDIATRIC

PATIENT DETAILS	
MRN:	Phone/ Mobile:
Surname:	Address:
Given Name: DOB:	
Gender: ☐ Female ☐ Male ☐ Unknown	Email:

PATIENT CONSENT

I understand:

- My child's DNA will be tested, by whole exome sequencing (WES), for genes associated with my child's condition.
- This test is NOT a general health test and will not identify all gene changes that could contribute to health problems in the future.
- Possible results: A range of clinical results may be reported. The results may include DNA variation that is well understood *or* results that are currently uncertain which may be clarified in the future or require further testing to interpret.
- There is a small chance genetic variants may be identified that are associated with an unrelated condition that may develop in the future, or that may reveal carrier status of an unrelated condition, these are defined as *incidental findings*. Incidental findings are rare (found in approximately 1% of cases). Only incidental findings that have a >90% confidence of being clinically relevant and are likely to develop in childhood are reported.
- Test results may have implications for the health care of my blood relatives.
- Testing may reveal non-paternity or non-maternity of a presumed natural parent.
- Testing will not currently affect the ability to obtain health insurance but may affect applications for some types of risk-rated insurances such as life and income protection insurance.
- My child's DNA sample and genomic data will be stored in accordance with national diagnostic laboratory guidelines.
- My child's genomic data and associated healthcare information can be used and disclosed in accordance with applicable health privacy laws.
- Testing is voluntary and I can withdraw or cancel testing at any stage.
- My child's de-identified genomic data and associated health information will be submitted to national or international clinical databases (restricted access).
- Sharing information with health practitioners involved in the care of the patient and genetic relatives is important in individual and family care. It reduces the work required for informing relevant practitioners and allows access to information that is relevant for other family members.

☐ **I consent** ☐ **I do not consent - to share my child's information with other relevant health practitioners.**

I consent to the genomic testing described above. Genomic testing has been explained to me by a health professional and I have had the opportunity to ask questions and I am satisfied with the explanations.

Patient / Parent / Guardian Name	Patient / Parent/ Guardian Signature	Date
Health Professional Name	Health Professional Signature	Date