

Test type	Recommended lab	Comments
Cardiac		
Cardiac Panels	1. Blueprint Genetics, Finland 2. Invitae, USA	
Aortopathy	1. Blueprint Genetics, Finland 2. Invitae, USA	PanelApp UK GMS Thoracic aortic aneurysm or dissection panel can be done at Canterbury Health laboratory, NZ
Endocrinology		
CaSR	Canterbury Health Laboratories, NZ	
Epigenetics		
11p Methylation/ Beckwith Wiedemann Syndrome	1. Monash Medical Centre, Australia 2. Institut für Humangenetik und Genommedizin (Thomas Eggermann), Germany	
Gastroenterology		
Chronic Pancreatitis Panel	1. Blueprint Genetics, Finland 2. Invitae, USA 3. Peter MacCallum Cancer Centre, Australia	Dosage and SNV 9 gene panel
Immunology		
Primary Immune Deficiency	1. Blueprint Genetics, Finland 2. Invitae, USA	
Bone Marrow Failure Panel	1. Blueprint Genetics, Finland 2. Invitae, USA	
Neurology		
Ataxia	CANVAS <i>RFC1</i> – University of Chicago, USA SCA – Concord, Australia or University of Chicago, USA 1. Blueprint Genetics, Finland 2. Invitae, USA 3. PathWest, Australia	SCA types 1, 2, 3, 6, 7 can be done at Wellington Regional Genetics Laboratory, NZ.
C9orf72	Wellington Regional Genetics Laboratory, NZ.	
Charcot Marie Tooth Neuropathy Panel	Blueprint Genetics, Finland	PMP22 can be done in-house.
Epilepsy	1. Blueprint Genetics, Finland 2. Invitae, USA	
FSHD	1. Bristol Genetics Laboratory, UK	*This testing requires a large volume of DNA. Therefore, 2x EDTAs must be collected to ensure there is enough or a recollect will be required.
Mitochondrial DNA Analysis	1. Canterbury Health Laboratories, NZ 2. Blueprint Genetics, Finland 3. PathWest, Australia	Nuclear 1.VCGS or 2.BpG only. VCGS has nuclear mitochondrial interpretation support for VOUS.
Myopathy Gene Panel	1. Blueprint Genetics, Finland 2. Invitae, USA 3. PathWest, Australia	Blueprint Genetics offers Comprehensive Muscular Dystrophy / Myopathy Panel.

Neuromuscular Gene Panel / Muscular NGS Panel	1. Blueprint Genetics, Finland 2. Invitae, USA 3. PathWest, Australia	Blueprint Genetics is always preferred. Blueprint Genetics offers Comprehensive Muscular Dystrophy / Myopathy Panel. PathWest don't report VOUS unless requested.
NF1/NF2	1. UAB Medical Genomics Laboratory, USA / Genomics for Life, Australia 2. Blueprint Genetics, Finland 3. Invitae, USA	Ideally if paired affected tissue with germline (lymphocytic DNA) – for number 1.
NOTCH3/CADASIL	Canterbury Health Laboratories, NZ	

Oncology

Fanconi Anaemia (Molecular Testing)	1. Blueprint Genetics, Finland 2. Invitae, USA	Cytogenetic test is done at Wellington Regional Genetics Laboratory, NZ.
Hereditary Cancer	1. Peter MacCallum Cancer Centre, Australia 2. Invitae, USA 3. Blueprint Genetics, Finland	For familial cases where positive controls were tested in another lab, sometimes testing will need to be sent to original lab.
Hereditary Paraganglioma-Pheochromocytoma Panel	1. Canterbury Health Laboratories, NZ 2. Blueprint Genetics, Finland 3. Invitae, USA	Single SDH gene testing can be done in-house. Panels will require sendaway.
Myeloid Gene Panel	1. Peter MacCallum Cancer Centre, Australia 2. Pathwest, Australia	

Skeletal Dysplasia

Ehlers-Danlos Syndrome Panel (Connective Tissue Disorder Panel/Collagenopathy)	1. Blueprint Genetics, Finland 2. Invitae, USA	PanelApp UK GMS Ehlers Danlos syndrome with a likely monogenic cause panel can be done at Canterbury Health laboratory, NZ
Overgrowth Syndrome / NDS1/Sotos Syndrome	1. Blueprint Genetics, Finland 2. Invitae, USA	Macrocephaly/overgrowth panel
Skeletal Dysplasia / Ciliopathies Panel/Osteogenesis Imperfecta Panel	1. Blueprint Genetics, Finland 2. Invitae, USA	PanelApp UK GMS Osteogenesis Imperfecta panel/ PanelApp UK GMS Skeletal dysplasia panel can be done at Canterbury Health laboratory, NZ

Other

Confirmatory Analysis/Custom Variant Analysis	Canterbury Health Laboratories, NZ	CHL will need to be contacted prior to sendaway request.
Macular Dystrophy panel	1. Blueprint Genetics, Finland 2. Invitae, USA 3. Molecular Vision Laboratories, USA	Vision panels can be done at MVL, USA.
Sensorineural Hearing Loss	1. Blueprint Genetics, Finland 2. Invitae, USA	
TSC1/TSC2 panel	1. Blueprint Genetics, Finland 2. Invitae, USA	
PanelApp Au RASopathy panel	Canterbury Health Laboratories, NZ	

UK GMS Rare syndromic craniosynostosis or isolated multisuture synostosis panel	Canterbury Health Laboratories, NZ	
WES	1. Blueprint Genetics, Finland 2. VCGS, Australia	Blueprint Genetics for all standard exomes – singleton, duo, trio, quad. VCGS can do Ultra rapid for urgent baby requests.
WGS (Trios)	Canterbury Health laboratory, NZ	

Note: The table above provides a simplified list of send-away tests available through our external laboratories.

If the test you require is not listed, please contact the relevant external laboratory directly for further assistance. Contact details for our main external laboratories are provided in resources.