Test type	Recommended lab	Comments
Cardiac		
Cardiac Panels	**1. Blueprint Genetics, Finland** 2. Invitae, USA	
Aortopathy PanelApp UK GMS Thoracic aortic aneurysm or dissection panel	**1. Canterbury Health Laboratories, NZ** 2. Blueprint Genetics, Finland 3. Invitae, USA	
Endocrinology		
CaSR	**Canterbury Health Laboratories, NZ**	
Epigenetics		
11p Methylation/ Beckwith Wiedemann Syndrome	**1. Institut für Humangenetik und Genommedizin (Thomas Eggermann), Germany** 2. Monash Medical Centre, Australia	Faster turnaround times with prompt responses to testing and result enquiries.
Gastroenterology		
Chronic Pancreatitis Panel	**1. Canterbury Health Laboratories, NZ** 2. Blueprint Genetics, Finland 3. Invitae, USA	
Immunology		
Primary Immune Deficiency	**1. Blueprint Genetics, Finland** 2. Invitae, USA	
Bone Marrow Failure Panel	**1. Blueprint Genetics, Finland** 2. Invitae, USA	
Neurology		
Ataxia	**Repeat Expansion testing** **1.Wellington Regional Genetics Laboratory, NZ** **2. University of Chicago, USA: Comprehensive repeat expansion testing** **NGS panels repeat expansions excluded** **1. Blueprint Genetics, Finland** 2. PathWest, Australia	First-line NZ testing for SCA types 1,2,3,6,7 Ataxia Repeat Expansion Panel including CANVAS/RFC1, SCA27B/FGF14, Friedreich ataxia, FXTAS and multiple SCAs
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. UKW, Germany** 2. Bristol Genetics Laboratory, UK – FSHD2 only currently	*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 offers Comprehensive Muscular Dystrophy / Myopathy Panel. PathWest don't report VOUS unless requested.
NF1/NF2	**1. Blueprint Genetics, Finland** 2. UAB Medical Genomics Laboratory, USA / Genomics for Life, Australia	UAB - Ideally if paired affected tissue with germline (lymphocytic DNA)
NF1	**Canterbury Health Laboratories, NZ**	
NOTCH3/CADASIL	**Canterbury Health Laboratories, NZ**	NOTCH3, HTRA1 genes
Oncology		
Fanconi Anaemia (Molecular Testing)	**1. Blueprint Genetics, Finland** 2. Invitae, USA	**Cytogenetic test (Chromosome Instability/Breakage Disorder)** 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. Canterbury Health Laboratories, NZ** 2. Blueprint Genetics, Finland	
Overgrowth Syndrome / NDS1/ Sotos Syndrome	**1. Blueprint Genetics, Finland** 2. Invitae, USA	Macrocephaly/overgrowth panel
Skeletal Dysplasia	**1. Canterbury Health Laboratories, NZ**	

PanelApp UK GMS Skeletal dysplasia panel	2. Blueprint Genetics, Finland	
Osteogenesis Imperfecta Panel PanelApp UK GMS Osteogenesis Imperfecta pane	**1. Canterbury Health Laboratories, NZ** 2. Blueprint Genetics, Finland	
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	
Genetic hearing loss gene panel	**Canterbury Health Laboratories, NZ**	GJB2, SLC26A4, GJB6 (del(GJB6-D13S1830), del(GJB6-D13S1854)
Comprehensive Sensorineural Hearing Loss	**1. Blueprint Genetics, Finland** 2. Invitae, USA	
TSC1/TSC2 panel	**1. Blueprint Genetics, Finland** 2. Invitae, USA	
Noonan syndrome PanelApp Au RASopathy panel	**Canterbury Health Laboratories, NZ**	
Craniosynostosis gene panel 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 (Trio)	**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.