The American Journal of Human Genetics, Volume *110*

Supplemental information

Australian Genomics: Outcomes of a 5-year national program to accelerate the integration of genomics in healthcare

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Table S1. Genomic testing approaches and diagnostic outcomes across each of the 13 rare disease and 6 cancer studies.

			Number of genomic tests				
Flagship study	**Participants recruited, number**	**Diagnosis/ Actionable result (%)**	**Panel**	**WES**	**WGS**	**Other***	**Relatives recruited, number**
Rare disease							
Acute Care Genomics	450	54	0	160	290	110	879
Neuromuscular disease	114	16	49	2	47	14	0
Mitochondrial disease	140	44	0	72	68	0	0
Epileptic encephalopathies	103	28	0	103	0	0	0
Brain malformations	102	44	0	102	0	0	0
Leukodystrophies	44	41	0	44	0	0	8
Intellectual disability	69	47	0	69	0	0	138
Genetic immune disorders	188	17	0	188	0	0	250
Perinatal genomic autopsy	202	28	0	194	0	0	620
Kidney genetics	357	43	74	210	51	22	0
Interstitial lung disease	35	17	0	30	5	0	63
Cardiovascular disorders	600	34	0	0	600	0	116
Unexplained end stage kidney disease	100	20	0	0	100	0	0
Cancer							
Acute lymphoblastic leukemia	374	100	0	0	0	519	0
Solid tumours (somatic)	368	63	308	0	0	0	0
Inherited cancer syndromes	195	7	0	0	195	0	19
Pediatric, adolescent and young adults	1659	6	0	0	1659	0	306
Lung cancer (somatic)	54	21	0	54	54	0	0

Cancers of unknown primary	118	92	92	0	30	78	0
TOTAL	5273		523	1228	3107	743	2399

*Transcriptome sequencing and multiplex ligation probe amplification (MLPA)