Table 1: Disease-associated markers in panel and corresponding pathogenicity thresholds.

Location (HG38)	**Gene**	**Disease**	**Pathogenicity threshold**	**OMIM link**
chr2:176093058-176093099	HOXD13	Synpolydactyly 1	21	186000
chr2:190880873-190880920	GLS	Global developmental delay, progressive ataxia, and elevated glutamine	90	618412
chr3:63912685-63912716	ATXN7	Spinocerebellar ataxia 7	37	164500
chr3:129172577-129172659	CNBP	Myotonic dystrophy 2	50	602668
chr4:3074877-3074940	HTT	Huntington disease	36	143100
chr4:41745976-41746022	PHOX2B	Congenital central hypoventilation	25	209880
chr5:146878728-146878759	PPP2R2B	Spinocerebellar ataxia 12	55	604326
chr6:16327634-16327724	ATXN1	Spinocerebellar ataxia 1	39	164400
chr6:45422750-45422794	RUNX2	Cleidocranial dysplasia	27	119600
chr6:170561907-170562017	TBP	Spinocerebellar ataxia 17	47	607136
chr7:27199680-27199729	HOXA13	Hand-foot-genital syndrome	18	140000
chr9:27573485-27573546	C9orf72	Amyotrophic lateral sclerosis	21	105550
chr9:69037285-69037304	FXN	Friedreich ataxia 1	200	229300
chr12:6936717-6936775	ATN1	Dentatorubral pallidoluysian atrophy	49	125370
chr12:111598950-111599019	ATXN2	Spinocerebellar ataxia 2	35	183090
chr13:70139384-70139429	ATXN8OS	Spinocerebellar ataxia 8	111	608768
chr13:99985449-99985494	ZIC2	Holoprosencephaly-5	25	609637
chr14:23321472-23321492	PABPN1	Oculopharyngeal muscular dystrophy	12	164300
chr14:92071011-92071052	ATXN3	Spinocerebellar ataxia 3	55	109150
chr16:87604283-87604329	JPH3	Huntington disease-like-2	50	606438
chr18:55586154-55586229	TCF4	Corneal dystrophy	40	613267
chr19:13207859-13207898	CACNA1A	Spinocerebellar ataxia 6	20	183086
chr19:18786035-18786050	COMP	Multiple epiphyseal dysplasia	6	132400
chr19:45770205-45770266	DMPK	Myotonic dystrophy 1	50	160900
chr20:2652733-2652775	NOP56	Spinocerebellar ataxia 36	650	614153
chr21:43776445-43776479	CSTB	Myoclonic epilepsy of Unverricht and Lundborg	30	254800
chr22:45795355-45795424	ATXN10	Spinocerebellar ataxia 10	800	603516