Gene	Chr**	Mode of inheritance	Disease	Comments
ABCD1*	X	X-linked	Adrenomyeloneuropathy	
AIFM1	X	X-linked	AIFM1 related disorders	
AR	X	X-linked	Androgen insensitivity syndrome	The current testing method does not assess CAG trinucleotide repeats in exon 1 in this gene.
CASK	X	X-linked	FG syndrome type 4; X-linked intellectual developmental disorder, Najm type	
CFTR	7	Autosomal recessive	Cystic fibrosis	Only variants associated with classical Cystic Fibrosis are reported. Intron 8 polymorphic region in CFTR gene (5T allele) is only reported when the NM_000492.4:c.350G>A (p.Arg117His) variant is detected.
CHM	X	X-linked	Choroideremia	
COL4A5	X	X-linked	Alport syndrome type 1	
DCX	X	X-linked	Lissencephaly type 1; Subcortical band heterotopia	
DMD	X	X-linked	Duchenne muscular dystrophy; Becker muscular dystrophy	
EDA	X	X-linked	Hypohidrotic ectodermal dysplasia type 1	
F8*	X	X-linked	Hemophilia A	Detection of intron 22 inversion in the F8 gene is also included
F9	X	X-linked	Hemophilia B	

Genes located in Chromosome X are only analysed in females.
*These genes have homology with other genomic loci and therefore the accuracy of the test may be decreased.
**Chromosome

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Gene	Chr**	Mode of inheritance	Disease	Comments
FMR1	X	X-linked	Fragile X syndrome	5' UTR CGG trinucleotide repeats are analysed. Only alleles in the range of full mutation (>200 repeats) and premutation (55-200 repeats) are reported. Mosaicism, including gonadal mosaicism, may not be detected.
FOXP3	X	X-linked	IPEX syndrome	
GJB2	13	Autosomal recessive	Deafness autosomal recessive type 1A	Variants associated with a mild phenotype are not reported.
GLA	X	X-linked	Fabry disease	
GPR143	X	X-linked	X-linked recessive ocular albinism; Congenital Nystagmus type 6	
HBA1-2	16	Autosomal recessive	Alpha-thalassemia	Whole gene analysis is not performed. Included variants: --MED ; --SEA ; --THAI ; - α3.7 ; - α4.2 ; - α20.5 ; --FIL; Hb Constant Spring (NM_000517.4:c.427T>C)
HBB	11	Autosomal recessive	Beta-thalassemia; Sickle cell disease	
IDS*	X	X-linked	Mucopolysaccharidosis type 2	
IQSEC2	X	X-linked	X-linked intellectual developmental disorder type 1	
MECP2	X	X-linked	RETT related disorders	
MTM1	X	X-linked	X-linked centronuclear myopathy	
OFD1	X	X-linked	Joubert syndrome type 10	

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Gene	Chr**	Mode of inheritance	Disease	Comments
OTC	X	X-linked	Ornithine transcarbamylase deficiency	
PRPS1*	X	X-linked	PRPS1 related disorders	
RS1	X	X-linked	Retinoschisis	
SLC6A8*	X	X-linked	Cerebral creatine deficiency syndrome type 1	
SMN1	5	Autosomal recessive	Spinal Muscular Atrophy	Only deletion of the exon 7 in SMN1 gene is included. No sequencing or deletion/duplication analysis is conducted in other regions of this gene. This test does not detect “silent” carriers of SMA, who have two copies of the SMN1 gene on one chromosome and none on the other.
SYN1	X	X-linked	X-linked epilepsy-learning disabilities-behavior disorders syndrome; X-linked intellectual developmental disorder type 50	
TAFAZZIN	X	X-linked	Barth syndrome	
WAS	X	X-linked	Wiskott-Aldrich syndrome; X-linked severe congenital neutropenia; X-linked thrombocytopenia	
XIAP*	X	X-linked	X-linked lymphoproliferative syndrome type 2	
ZIC3	X	X-linked	X-linked heterotaxia; VACTERL with hydrocephalus	

Genes located in Chromosome X are only analysed in females.
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**Chromosome

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