List of Xcelom Send Out Test - LRS

Service	Testing Scope	Sample Type	TAT
Thalassemia	**637 Panel** Covers 637 variants associated with α-thalassemia, β-thalassemia, and hemoglobinopathies (including sickle cell anemia), including: • *HBA1* and *HBA2* genes: 30 deletions, 4 homologous recombination, and 219 SNVs/InDels • *HBB* gene: 28 deletions, and 356 SNVs/InDels	Peripheral blood, DBS, or gDNA	15 working days
	Expanded Panel Covers 2025 variants associated with α-thalassemia, β-thalassemia, hemoglobinopathies, and hemoglobin variants in the *HBA1, HBA2,* and *HBB* genes, including: • *HBA1* and *HBA2* genes: 30 deletions, 4 homologous recombination, and 929 SNVs/InDels • *HBB* gene: 28 deletions, and 1034 SNVs/InDels		
	Comprehensive Panel Covers 2,288 variants, including 2,026 variants associated with α-thalassemia, β-thalassemia, hemoglobinopathies, and hemoglobin variants. It also includes 160 SNVs/InDels related to the *HBD* gene and 102 SNVs/InDels in the *HBA* and *HBB* genes that are low frequency, not clearly associated with thalassemia or hemoglobin abnormalities		
	Prenatal Comprehensive Panel Targets for the genes and variants related to related to thalassemia, such as *HBA1*, *HBA2*, and *HBB*, including P/LP SNVs/InDels associated with α-thalassemia, β-thalassemia, hemoglobinopathies, and hemoglobin variants. It also includes P/LP SNVs/InDels associated with the *HBD* gene, as well as *HBA* and *HBB* gene mutations that are low frequency, not clearly associated with thalassemia or hemoglobin abnormalities	Prenatal sample: gDNA from amniotic fluid Maternal sample: Peripheral blood or gDNA	
Spinal Muscular Atrophy	**Basic Panel** Detects *SMN1* and *SMN2* full-length copy number, and 133 P/LP *SMN1* variants	Peripheral blood, DBS, or gDNA	15 working days
	Comprehensive Panel Detects *SMN1* and *SMN2* full-length copy number, and a total of 188 *SMN1* variants classified as P/LP/VUS		
	Trio [2+0] Carrier Add-on Determines whether the individual is a silent [2+0] carrier by analyzing the *SMN1* full-length copy number in family relatives and confirming the haplotype through family-based analysis		
Congenital Adrenal Hyperplasia	**Basic Panel** Detects 514 P/LP variants, including: • *CYP21A2-TNXB* genes: 12 deletions, 273 SNVs/InDels, and 12 CNVs • *CYP11B1* gene: 1 deletion and 79 SNVs/InDels • *CYP17A1* gene: 55 SNVs/InDels • *HSD3B2* gene: 36 SNVs/InDels • *STAR* gene: 46 SNVs/InDels	Peripheral blood, DBS, or gDNA	15 working days
	Comprehensive Panel Detects P, LP, and some VUS SNVs/InDels, selected large intragenic deletions in 7 genes (*CYP21A2*, *CYP11B1*, *CYP17A1*, *HSD3B2*, *STAR*, *POR*, and *CYP11A1*), and specific CNVs in the *CYP21A2* gene		
	Prenatal Comprehensive Panel Detects P/LP SNVs/InDels and selected large intragenic deletions in 7 genes (*CYP21A2*, *CYP11B1*, *CYP17A1*, *HSD3B2*, *STAR*, *POR*, and *CYP11A1*), and specific CNVs in the *CYP21A2* gene	Prenatal sample: gDNA from amniotic fluid Maternal sample: Peripheral blood or gDNA	

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Fragile X Syndrome	**Basic Panel** Determines the number of CGG repeats in the 5' UTR region of the *FMR1* gene, and AGG interruption numbers and positions	Peripheral blood, DBS, or gDNA	15 working days
	Comprehensive Panel Determines the number of CGG repeats in the 5' UTR region of the *FMR1* gene, AGG interruption numbers and positions, and exon 1 deletions		
	Prenatal Comprehensive Panel Determines the number of CGG repeats in the 5' UTR region of the *FMR1* gene, AGG interruption numbers and positions, and exon 1 deletions	Prenatal sample: gDNA from amniotic fluid Maternal sample: Peripheral blood or gDNA	
Hemophilia	**Basic panel** Detects *F8* gene intron 1/22 inversions and exon 22 deletions/duplications	Peripheral blood, DBS, or gDNA	17 working days
	Comprehensive panel Detects hemophilia related variants, including: • *F8* gene: Intron 1/22 inversions, exon 22 deletions/duplications, P/LP/selected VUS SNVs/InDels, and selected P/LP/VUS large deletions/duplications • *F9* gene: P/LP/selected VUS SNVs/InDels, and selected P/LP/VUS large deletions/duplications • *VWF* gene: Potentially clinically significant SNVs/InDels SNVs/InDels in exons 18-28 (the region responsible for binding factor VIII (F8))		
	Prenatal Comprehensive Panel Detects hemophilia related variants, including: • *F8* gene: Intron 1/22 inversions, exon 22 deletions/duplications, P/LP SNVs/InDels, and selected P/LP large deletions/duplications • *F9* gene: P/LP SNVs/InDels, and selected P/LP large deletions/duplications • *VWF* gene: P/LP SNVs/InDels in exons 18-28 (the region responsible for binding factor VIII (F8))	Prenatal sample: gDNA from amniotic fluid Maternal sample: Peripheral blood or gDNA	
Autosomal Dominant Polycystic Kidney Disease	Detects autosomal dominant polycystic kidney disease-associated variants in the *PKD1* and *PKD2* genes, including: • P, LP, and some VUS SNVs/InDels • Exon deletions/duplications	Peripheral blood, DBS, or gDNA	17 working days
Fabry Disease	Detects the *GLA* variants associated with Fabry disease, including: • P, LP, and some VUS SNVs/InDels • Some large intragenic deletions/duplications classified as P/LP/VUS	Peripheral blood, DBS, or gDNA	17 working days
Hunter Syndrome (Mucopolysaccharidosis II)	Detects the *IDS* variants associated with mucopolysaccharidosis II, including: • P, LP, and some VUS SNVs/InDels • Some large intragenic deletions/duplications classified as P/LP/VUS • Intron 7 inversion • Exon 2-intron 3 inversion	Peripheral blood, DBS, or gDNA	17 working days

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Incontinentia Pigmenti	Detects the *IKBKG* variants associated with Incontinentia Pigmenti, including: • P, LP, and some VUS SNVs/InDels • Some large intragenic deletions classified as P/LP/VUS • Some complex rearrangements	Peripheral blood, DBS, or gDNA	15 working days
Gaucher disease	Detects the *GBA1* variants associated with Gaucher disease, including: • P, LP, and some VUS SNVs/InDels • Some large intragenic deletions classified as P/LP/VUS	Peripheral blood, DBS, or gDNA	15 working days
Ichthyosis Vulgaris	Detects the *FLG* variants associated with Ichthyosis Vulgaris, including: P, LP, and some VUS SNVs/InDels within exon 3	Peripheral blood, DBS, or gDNA	15 working days
Newborn genetic screening (LRS)	• 21-hydroxylase deficiency: 316 P/LP variants in the *CYP21A2* gene, including 12 deletions, 292 SNVs/InDels, and 12 CNVs • Non-syndromic hearing loss: 262 P/LP variants in the *SLC26A4*, *GJB2*, *GJB3*, and *MT-RNR1* genes, including 146 SNVs/InDels in *SLC26A4* gene, 109 SNVs/InDels in *GJB2* gene, 4 SNVs/InDels in *GJB3* gene, and 3 SNVs/InDels in *MT-RNR1* gene • Phenylketonuria and BH4-deficient HPA: 485 P/LP SNVs/InDels in the *PAH* gene, and 19 P/LP SNVs/InDels in the *PTS* gene • Glucose-6-phosphate dehydrogenase deficiency: 69 P/LP SNVs/InDels in the *G6PD* gene	Peripheral blood, DBS, or gDNA	15 working days
dmTGS (Massive Tandem Repeat Panel)	Analyzes 63 genes and 68 diseases associated with dynamic mutations, detecting repeat numbers related to these disorders	Peripheral blood or gDNA	29 working days
HiFi LR-WGS (HiFi Long-Read Whole Genome Sequencing)	Reporting variants: SNVs, InDels ≤50 bp, CNVs, structural variants, absence of heterozygosity (AOH)/ uniparental disomy (UPD, for trio), mitochondrial variants ≤50bp, tandem repeat/dynamic variants in 63 genes, and abnormal methylation for 8 diseases	Peripheral blood or gDNA	34 working days

Test customization is available.
For details, please contact **marketing@xcelom.com**

Note:
TAT starts from the date of sample arrival at the testing laboratory.
It is recommended to provide the maternal sample along with the prenatal sample to rule out maternal contamination.
Sample QC and transportation requirements vary by test. Please contact us for details.

Website (Global) **LinkedIn**

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