List of Xcelom Send Out Test - NGS

Service	Testing Scope	Sample Type	TAT
Non-Invasive Prenatal Testing (NIPT) Illumina NGS	Fetal chromosomal aneuploidies caused by trisomies, SCA and microdeletions >3Mb, as well as other chromosomal and sub-chromosomal findings >3Mb	Maternal blood (≥ 10 weeks for singleton, ≥ 12 weeks for twin, and ≥ 14 weeks for triplet)	5 working days
Non-Invasive Prenatal Testing of Single Gene Disorders (NIPT-SGD) Illumina NGS	P/LP SNVs/InDels ≤3bp within 34 genes associated with 66 single gene disorders Note: P/LP InDels >3bp within 34 genes associated with the 66 disorders is reported as supplementary	Maternal blood (≥ 10 weeks for singleton pregnancy only)	20 working days
CNV-seq CNVs: Illumina NGS UPD add-on: Fluorescent PCR-capillary electrophoresis	Genetic disorders caused by chromosomal aneuploidies or CNVs > 100 kb with mosaicism UPD add-on testing for 10 UPD-related disorders available with prenatal CNV-seq	Peripheral blood, cord blood, or gDNA	14 working days
PGT-A Illumina NGS	Aneuploidies and microdeletions/microduplications >10Mb with mosaicism	Trophoblasts with blank control	14 working days
smrtPGT-M/-SR, PGT-M/-SR PGT-M/-SR: Illumina Infinium BeadChip smrtPGT-M/-SR: PacBio SMRT sequencing	**PGT-M** Known single gene disorders, HLA typing, and genetic susceptibility-related tumors (36 disorders) **PGT-SR** Balanced translocations, Robertsonian translocations, inversions, and pathogenic microdeletions/microduplications <10Mb **smrtPGT-M/-SR + PGT-M/-SR** Known single gene disorders/ Balanced translocations and inversions	Embryo: Trophoblasts with blank control; Parent/ Family reference: Peripheral blood or gDNA	PGT-M/-SR: 14 working days smrtPGT-M/-SR + PGT-M/-SR: 21 working days
Expanded Carrier Screening Illumina NGS	**21-Gene Panel** Detects 4,032 P or LP variants across 21 genes **51-Gene Panel** Detects 7,343 P or LP variants across 51 genes **155-Disorders Panel** Detects 11,280 P/LP variants in 129 genes associated with 155 X-linked and autosomal recessive disorders **1784-Disorders Panel** Screens for 1,784 autosomal recessive and X-linked recessive disorders associated with 1,766 genes (This panel is available only for couple-based testing. We report P/LP variants only when both partners carry variants in the same gene, or when the female partner carries an X-linked variant. Single-carrier findings for autosomal recessive conditions and VUS are not disclosed in the final report.) Variant Types Reported: SNVs in exons and splice boundaries, InDels < 50bp in exons, and selected CNVs in *DMD*, *SMN1*, and *HBA1/2* genes (if included in panel). Add-on analysis with LRS is available for Fragile X Syndrome (CGG/AGG repeats) and hemophilia A (*F8* Inv1/ 22). **Add-on:** Fragile X syndrome - CGG repeat, Hemophilia A - Inversion of introns 1 and 22 in the *F8* gene	Peripheral blood or gDNA	20 working days

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Newborn Genetic Screening **-** **NGS approach** Illumina NGS	**Jaundice Panel** Detects P/LP variants in 82 genes associated with single-gene disorders presenting with a jaundice phenotype **Inherited Metabolic Disorders Panel** Detects P/LP variants in 57 genes associated with 72 inherited metabolic diseases **411-Disorder Panel** Detects P/LP variants across 358 target genes associated with 411 single-gene disorders **1264-Disorder Panel** Detects P/LP variants across 737 genes associated with 1,264 single-gene disorders Reporting: P and LP variants (SNVs in exons and ±5 bp exon–intron boundaries; InDels <50bp in exon; selected CNVs in *DMD*, *SMN1*, or *HBA1/2* genes (if included in panel)) within the scope are reported. Carrier status for autosomal recessive and X-linked recessive conditions is not reported.	Peripheral blood, DBS, or gDNA	15 working days
Prenatal / General Whole Exome Sequencing (WES) Illumina NGS	Targeted regions: Genes with defined molecular mechanisms in OMIM, expert-reviewed genes in ClinGen, regulatory elements/ intronic regions with defined pathogenicity, mitochondrial genome, and RNA regions in RefGene Type of variants reported: SNVs, InDels, exonic CNVs, aneuploidy, loss of heterozygosity (LOH)/ uniparental disomy (UPD, for trio), and dynamic mutations (trinucleotide repeats) in clinically well-known genes	Peripheral blood, DBS, cord blood, or gDNA	Prenatal WES: 15 working days General WES: 23 working days
Whole Genome Sequencing (WGS) Illumina NGS	Type of variants: SNVs, InDels, CNVs, aneuploidy, loss of heterozygosity (LOH)/ uniparental disomy (UPD, for trio), and dynamic mutations (trinucleotide repeats) in clinically well-known genes	Peripheral blood, cord 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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