cell genetics Laboratory

BabyGenome

Your baby's genetic passport

welcome baby!

BabyGenome
Your baby's genetic passport

is a **clinical genetic test** specifically designed for **newborns** and **children under 3 years of age**, which aims to exclude the presence of pathogenic and likely pathogenic sequence variants in **106 genes** associated with **rare, childhood-onset genetic disorders** for which **early medical intervention is available** allowing for effective management of the condition and alleviation of corresponding symptoms.

The DNA test BabyGenome covers 106 genes associated with various conditions which were **recommended for uniform newborn genetic screening by the American College of Obstetricians and Gynecologists** (ACOG). For the purpose of this genetic screen**, whole-genome sequencing (WGS) is performed** which uncovers close to a 100% of the DNA sequence within the human nuclear and mitochondrial genome.

List of the tested genetic conditions:

Inborn errors of organic acid metabolism:

- ✓ Isovaleric acidaemia
- ✓ 3-Methylcrotonyl-CoA carboxylase deficiency, type I, II
- ✓ Methylmalonic acidaemia (cobalamin disorders)
- ✓ Methylmalonic acidaemia (methylmalonyl-CoA mutase)
- ✓ β-ketothiolase deficiency
- ✓ Glutaric acidaemia, type I
- ✓ Propionic acidaemia
- ✓ Holocarboxylase synthetase deficiency
- ✓ 3-Hydroxy-3-methylglutaric aciduria

Inborn errors of fatty acid metabolism:

- ✓ Medium-chain acyl-CoA dehydrogenase deficiency
- ✓ Very long-chain acyl-CoA dehydrogenase deficiency
- ✓ Long-chain L-3 hydroxyacyl-CoA dehydrogenase deficiency
- ✓ Trifunctional protein deficiency
- ✓ Carnitine uptake defect/transport defect

Inborn errors of amino acid metabolism:

- ✓ Classic phenylketonuria
- ✓ Maple syrup urine disease
- ✓ Homocystinuria
- ✓ Citrullinemia, type I
- ✓ Argininosuccinic aciduria
- ✓ Tyrosinemia, type I

Haemoglobinopathies:

- ✓ Beta thalassemia
- ✓ Sickle cell anemia

Miscellaneous multisystem diseases:

- ✓ Biotinidase deficiency
- ✓ Congenital adrenal hyperplasia
- ✓ Primary congenital hypothyroidism
- ✓ Classic galactosaemia
- ✓ Cystic fibrosis
- ✓ Glycogen Storage Disease Type II (Pompe)
- ✓ Mucopolysaccharidosis, type I
- ✓ Severe combined immunodeficiency
- ✓ X-linked adrenoleukodystrophy

List of the genes analyzed:

ABCD1	*BCL10*	*CORO1A*	*FAH*	*HMGCL*	*LAT*	*MMUT*	*PPM1K*	*SLC5A5*	*TRAC*
ABCD4	*BTD*	*CYP11A1*	*FOXN1*	*HSD3B2*	*LCK*	*MSN*	*PRDX1*	*STAR*	*TRHR*
ACADM	*CBS*	*CYP11B1*	*GAA*	*IDUA*	*LIG4*	*MTHFR*	*PRKDC*	*STAT3*	*TSHB*
ACADVL	*CD247*	*CYP17A1*	*GALT*	*IL2RG*	*LMBRD1*	*NBN*	*PTPRC*	*STK4*	*TSHR*
ACAT1	*CD3D*	*CYP21A2*	*GCDH*	*IL7R*	*MALT1*	*NHEJ1*	*RAG1*	*TAPBP*	*TTC7A*
ADA	*CD3E*	*DBT*	*HADH*	*IRS4*	*MCCC1*	*NKX2-5*	*RAG2*	*TBL1X*	*WAS*
AK2	*CD3G*	*DCLRE1C*	*HADHA*	*IVD*	*MCCC2*	*PAH*	*RFXANK*	*TFRC*	*ZAP70*
ASL	*CD40LG*	*DLD*	*HADHB*	*IYD*	*MMAA*	*PAX8*	*RMRP*	*TG*	
ASS1	*CD8A*	*DOCK2*	*HBB*	*JAK3*	*MMAB*	*PCCA*	*RNF168*	*THRA*	
BCKDHA	*CFTR*	*DUOX2*	*HCFC1*	*KDM6A*	*MMACHC*	*PCCB*	*SLC22A5*	*THRB*	
BCKDHB	*CIITA*	*DUOXA2*	*HLCS*	*KMT2D*	*MMADHC*	*POR*	*SLC26A4*	*TPO*	

***The list of genetic conditions was developed following the recommendations from ACOG Committee Opinion No. 778**: Newborn Screening and the Role of the Obstetrician-Gynecologist. Obstet Gynecol. 2019. May;133(5):e357-e361. PMID: 31022124*

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