Picture™

FOR CLINICIANS

Picture Newborn Information Sheet

Who is Picture Genetics?

Picture Genetics puts patients first by providing at-home, clinical-grade genetic testing products paired with certified genetic counselor guidance. Our test results are designed to be straightforward and clinically actionable. Analysis for Picture Genetics tests is performed at Fulgent Genetics, a CLIA-certified and CAP-accredited diagnostic laboratory working with physicians from more than 700 institutions around the world.

What is the Picture Newborn test?

Picture Newborn analyzes **257 genes** that are associated with early-onset conditions that can be treated if identified early in life. It is designed to complement and enhance state-mandated standard newborn screening by analyzing more conditions and providing more certainty.

- Picture Newborn screens for over 200 rare genetic conditions, compared to an average of 30 conditions for standard state screening.
- Picture Newborn is a diagnostic test, reducing the need for follow-up testing.
- Picture Newborn requires just a simple and gentle cheek swab for analysis (no "heel prick").

For more information on state-mandated Newborn Screening, please visit https://www.babysfirsttest.org/

Who should order this test?

Picture Newborn is for parents who are interested in determining whether their child is at risk of developing certain severe or life-altering genetic conditions, regardless of family history. The test looks for potential conditions with symptoms that would appear before the age of 10. The majority of these conditions usually show symptoms before the age of one.

What kinds of conditions does Picture Newborn test for?

Picture Newborn looks for conditions that could seriously impact a child's quality of life, including childhood cancers, epilepsy, cardiovascular disorders, and hearing and vision loss. All of these conditions are considered rare, and all are associated with a form of treatment that can make a significant positive difference in a child's health when identified early. For a full list of the genes and conditions that Picture Newborn tests for, see below.

What benefits can this test provide?

Aside from early diagnosis and early intervention, positive results may mean that other family members carry the same genetic change and could benefit from genetic testing. A positive result may also mean that any future children could be affected by the same genetic condition. It's recommended that patients discuss this with a genetic counselor (included in the cost) to get the most informed view of these implications in the context of their unique family history.

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Can insurance help pay for this test?

Picture Genetics currently does not support insurance payments for our tests.

A snapshot of test specifications

Coverage ~99% at 20x **Turnaround Time** 3-5 weeks **Specimen Type** Saliva

Genes Tested 257 total | Metabolic (148), Blood (12), Hearing Loss (18), Cardiac (8), Immunodeficiency (22), Pediatric Cancer (13), Epilepsy (10), Vision Loss (4), Other (22)

Analysis Sequencing & Deletion/Duplication

Reporting

Only variants classified as "Pathogenic" or "Likely Pathogenic" using the ACMG guidelines for sequence variant interpretation will be reported. We will not report Variants of Uncertain Significance (VUS) or carrier status.

Detection rate

A broad range of laboratory and bioinformatic tools are employed to ensure the highest detection rate. Picture Newborn's analytical detection rate for all genes is >98%.

Test methods

Next generation sequencing and deletion/duplication analysis
Sequencing reads the DNA code of the selected genes, one base at a time, to determine an individual's sequence. The sequence is then compared to a reference DNA sample to detect any variants found within the patient's DNA. Next generation sequencing (NGS) is used to analyze exons in multiple genes simultaneously. Picture Genetics uses a sophisticated method that detects sequence changes and deletions/duplications (del/dups) via NGS. Pathogenic variants found by this method are confirmed by Sanger sequencing, MLPA, or quantitative PCR (qPCR).

Test limitations

All laboratory tests have limitations. A positive result does not imply that there are no other mutations in the patient's genome, and negative results do not eliminate the risk for the patient to be affected by a genetic disorder. Picture Newborn is not designed to detect somatic mutations. Mutations that are not located in the exons of genes may not be detected by this test.

If you have questions, feel free to call or email us.

PICTURE NEWBORN

Genes and Conditions Tested

Conditions & Genes Associated

Metabolic Disorders

ABCC8, ABCD1, ABCG5, ACAD8, ACADM, ACADVL, ACAT1, AGL, AGXT, AKR1D1, ALDOB, ALPL, AQP2, ARG1, ARSA, ARSB, ASL, ASS1, ATP7A, ATP7B, AVPR2, BCKDHA, BCKDHB, BTD, CASR, CBS, CPS1, CPT1A, CPT2, CTNS, CYP11B1, CYP11B2, CYP17A1, CYP27A1, CYP27B1, DBT, DLD, DUOX2, DUOXA2, ETFA, ETFB, ETFDH, FAH, FBP1, FOLR1, G6PC, G6PD, GAA, GALC, GALE, GALK1, GALNS, GALT, GAMT, GATM, GBA, GCDH, GLA, GLB1, GLUD1, GRHPR, GSS, GUSB, GYS2, HADH, HADHA, HADHB, HLCS, HMGCL, HMGCS2, HNF4A, HOGA1, HPD, HSD3B2, HSD3B7, IDS, IDUA, IGSF1, INS, IVD, IYD, KCNJ11, LDLR, LHX3, LIPA, LMBRD1, LPL, MAT1A, MC2R, MCCC1, MCCC2, MCEE, MLYCD, MMAA, MMAB, MMACHC, MMADHC, MPI, MTR, MTRR, MTTP, MUT, NAGS, NPC1, NR0B1, OTC, PAH, PAX8, PCBD1, PCCA, PCCB, PHEX, PHGDH, PKLR, POR, POU1F1, PROP1, PTS, PYGL, QDPR, SCNN1A, SCNN1B, SLC22A5, SLC25A13, SLC25A15, SLC25A20, SLC2A1, SLC34A3, SLC37A4, SLC39A4, SLC5A5, SLC7A7, SMPD1, SPR, STAR, SURF1, TAT, TCN2, TG, TH, THRA, TPO, TRHR, TRMU, TSHB, TSHR, UGT1A1, VDR

Blood Disorders

ACVRL1, ADAMTS13, ANK1, ELANE, ENG, EPB42, F9, HAX1, HBB, MPL, SLC4A1, SPTB

Hearing Loss

CDH23, GJB2, GJB6, KCNE1, MYO15A, OTOF, PAX3, SLC26A4, SOX10, TECTA, TMIE, TMPRSS3, TPRN, TRIOBP, USH1C, USH1G, USH2A, WHRN

Cardiac Conditions

FBN1, LAMP2, PTPN11, SMAD3, TAZ, TGFBR1, TGFBR2, ZIC3

Immunodeficiency Disorders (SCID)

ADA, BTK, CD3D, CD3E, CD40LG, CIITA, CYBA, CYBB, DCLRE1C, DOCK8, FOXP3, IL2RG, IL7R, JAK3, NCF2, PTPRC, RAG1, RAG2, RFX5, RFXANK, RFXAP, ZAP70

Pediatric Cancers

BMPR1A, ERCC2, ERCC5, MEN1, NF1, NF2, PTCH1, RB1, SMAD4, STK11, VHL, XPA, XPC

Epilepsy

ALDH7A1, ETHE1, KCNQ2, PNPO, PRRT2, SCN1A, SCN2A, SCN8A, TSC1, TSC2

Vision Loss

OAT, OPA1, GPR143, SLC45A2

Other Conditions

ANKH, CFTR, COL1A1, COL1A2, COL4A3, COL4A4, COL4A5, CRLF1, GCH1, HPS1, HPS4, JAG1, MEFV, NTRK1, PKD2, PKHD1, PRF1, SMN1, SMN2, TCIRG1, TTPA, WT1

