Picture™

FOR CLINICIANS

Picture Parenting 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 Parenting test?

Picture Parenting is a physician-backed, patient-oriented carrier screening test. Carrier screening uses genetic testing to identify couples and individuals at risk of passing genetic conditions on to their children. Picture Parenting is unique in that it can be easily ordered and completed from home, but still includes physician involvement from an independent physician network (PWNHealth) and genetic counseling support for your patient.

Who should order this test?

Carrier screening is ideal for individuals who are currently pregnant or are considering pregnancy. The American College of Obstetricians and Gynecologists (ACOG) recommends that pregnant women and couples who are considering pregnancy have information about and access to carrier screening.

What are the benefits this test can provide?

Understanding your patient's carrier status allows you and your patient to work together to discuss reproductive planning options best suited for their particular journey. Couples who are found to be carriers for the same condition generally have a 25% chance of having a child affected with that condition. Carrier couples who are not yet pregnant may wish to discuss options such as in vitro fertilization (IVF) with preimplantation genetic testing, gamete donors, or adoption; while carriers who are currently pregnant can consider additional prenatal testing and physician guidance.

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 30 (female) or 28 (male)
SMA Possible "Silent Carrier" status is reported with population-specific variants
Fragile X AGG interruptions analyzed and reported for premutation carriers

FOR CLINICIANS

Picture Parenting Information Sheet

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).

Detection rate

A broad range of laboratory and bioinformatic tools are employed to ensure the highest detection rate. Picture Parenting'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. This helps ensure that we can identify carriers for conditions such as spinal muscular atrophy (SMA). Pathogenic variants found by this method are confirmed by Sanger sequencing, MLPA, or quantitative PCR (qPCR).

Pseudogene interpretation
Sometimes genetic material that resembles a real gene (pseudogene) or genes that contain similar sequences may interfere with the ability to identify mutations via NGS. To bypass this issue, Picture Genetics uses highly sensitive tools that are capable of identifying carrier mutations in disease genes (such as GBA for Gaucher disease and HBA1/HBA2 for alpha-thalassemia) without the risk of pseudogene interference.

PCR amplification
PCR amplification is used to detect the CGG repeat expansion of the FMR1 gene. When the CGG repeat expands to a specific number, it can cause Fragile X syndrome. We can also detect AGG interruptions, which may decrease the risk of the CGG repeat expansion when inherited from the mother.

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's children to be affected with a genetic disorder. Picture Parenting is not designed to detect somatic mutations. Mutations that are not located in the exons of genes may not be detected by this test. For gene specific limitations, please email us.

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

PICTURE PARENTING

Genes and Conditions Tested

Gene / Condition Name

***ACADM* -** Medium-chain acyl-CoA dehydrogenase (MCAD) deficiency
***ASPA* -** Canavan disease
***ASS1* -** Citrullinemia
***BLM* -** Bloom syndrome
***CFTR* -** Cystic fibrosis
***CLN3* -** Neuronal ceroid lipofuscinosis, CLN3-related
***DHCR7* -** Smith-Lemli-Opitz syndrome
***DMD** -** Duchenne muscular dystrophy
***FANCC* -** Fanconi anemia group C
***FAH* -** Tyrosinemia, type 1
***FMR1** -** Fragile X syndrome
***G6PC* -** Glycogen storage disease, type 1a
***GAA* -** Pompe disease
***GALT* -** Galactosemia
***GBA* -** Gaucher disease
***HBA1 / HBA2* -** Alpha thalassemia
***HBB* -** Sickle cell disease; beta thalassemia
***HEXA* -** Tay-Sachs disease
***IDUA* -** Mucopolysaccharidosis, type I (Hurler syndrome)
***PAH* -** Phenylalanine hydroxylase deficiency (Phenylketonuria)
***IVD* -** Isovaleric acidemia
***PEX1* -** Zellweger syndrome, PEX1-related
***PEX7* -** Rhizomelic chondrodysplasia punctata, type 1
***PKHD1* -** Polycystic kidney disease, PKHD1-related
***PMM2* -** Congenital disorder of glycosylation type 1a
***SMN1* -** Spinal muscular atrophy
***SMPD1* -** Niemann-Pick disease, type A/B
***IKBKAP* -** Familial dysautonomia
***MMACHC* -** Methylmalonic aciduria and homocystinuria, cblC type

* Male patients will not be screened for X-linked conditions. If an X-linked condition is suspected in a male patient, please contact a genetics professional about diagnostic testing for that particular disorder.

