

Prenatal Diagnosis

Price change effective August 1, 2008 Prenatal diagnosis for known mutations now \$2000
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GeneDx can provide prenatal diagnosis for known familial mutations, in any gene, for families that have had previous testing at GeneDx or elsewhere. For genes not on GeneDx's standard menu, early preparation is critical. If all preliminary testing of controls or parents is complete, the turnaround time when testing known familial mutations is usually 2 weeks. Call 301-519-2100 or email genedx@genedx.com to speak to a genetic counselor in advance.

Prenatal Diagnosis for Known Familial Mutations includes

- Case-management by a genetic counselor
- Duplicate analysis of fetal specimen
- Maternal cell contamination studies
- Comparison to positive familial controls (required)
- Usually 2-week turnaround if GeneDx has already tested key family members

Choose one of the following links for specific step-by-step instructions

[Known Familial Mutations in Autosomal Recessive Disorders](#)

[Known Familial Mutations in Autosomal Dominant Disorders](#)

[Known Familial Mutations in X-Linked Disorders](#)

In addition, for a limited number of genes we offer full gene sequencing based on fetal ultrasound indications.

- Examples of disorders that may present for full gene sequencing with fetal ultrasound findings and without known familial mutations are Noonan Syndrome, Androgen Insensitivity, Smith-Lemli-Opitz Syndrome, Holoprosencephaly, Holt-Oram Syndrome, Townes-Brocks Syndrome, Duane-RadialRay/Acro-Renal-Ocular Syndrome, and X-linked hydrocephalus.
- For more information for each test, see the printable Information Sheet under the Gene or Disorder on our menu of Diagnostic Tests. For Noonan Syndrome, on the Disorder menu see the separate link for Noonan Prenatal Testing.
- Maternal blood (3 mL in EDTA) is required for concurrent maternal cell contamination (MCC) studies
- Sequencing will be rushed for ongoing pregnancies. The turnaround time will be faster than for pediatric cases but nevertheless may be several weeks.
- For specimen requirements see [Amnio/CVS/Cultures](#) under Send a Specimen
- Call 301-519-2100 or email genedx@genedx.com to speak to a genetic counselor in advance.