

Prenatal Diagnosis for Known Familial Mutations in Autosomal Recessive Disorders

For recessive disorders, testing of both parents is an essential aspect of accurate prenatal diagnosis.

If the proband was diagnosed at GeneDx, the next step is parental carrier testing. After that, subsequent prenatal diagnosis usually can be completed with a 2-week turnaround. If more than 6 months have elapsed since Carrier Testing, fresh parental specimens should accompany the fetal specimen for control purposes as needed in gene-specific and MCC tests.

If the proband was tested in another CLIA laboratory or a research laboratory, see the corresponding column below. If parents do not order Carrier Tests at GeneDx in advance, the 2-week TAT for prenatal diagnosis does not apply. If outside testing was done in a research laboratory, advance parental Carrier Tests are mandatory.

STEPS for Recessive Disorders		Previous Testing Performed By		
		GeneDx	Other CLIA Laboratory	Research Laboratory
1	Submit the outside molecular lab results to GeneDx for review and pre-approval	Not necessary	Required	Required
2	If possible provide DNA from the proband as a control*	Not necessary	Recommended*	Strongly Recommended*
3	Order Parental carrier testing (#9011 or #9012 below) for each parent, ideally 1 month or more in advance.	Strongly Recommended if not already done at GeneDx	Strongly Recommended if not already done at GeneDx	Required if not already done at GeneDx
4	Submit a fetal specimen for prenatal diagnosis (#902 below). Parental specimens must accompany the fetal specimen, if not sent within 6 months, even if parental carrier tests are not ordered.	TAT will be 2 weeks in most cases	TAT will be 2 weeks in most cases, if Step 3 is already complete	TAT will be 2 weeks in most cases, if Step 3 is already complete

*If a Mutation Confirmation service with a report is desired on a proband to confirm a research result, send a fresh blood or buccal specimen and order 9001 or 9002 as below.

9001	Mutation Confirmation for research results, one mutation, heterozygous or homozygous. \$350. For independent confirmation send fresh blood, 1-3 mL in EDTA, or a GeneDx buccal kit. If a fresh specimen is not available, 5 mcg stored DNA will be accepted. An individual report is provided. TAT 4 weeks. For CPT codes see the Information Sheet under Mutation Confirmation on the main web menu.
9002	Mutation Confirmation as above, for two distinct mutations. \$500.
9011	Carrier Testing for one familial mutation. \$350. Specimens can be 1-3 mL EDTA blood, a GeneDx buccal kit, or 5 mcg DNA. An individual report is provided. TAT 4 weeks. For CPT codes see the Information Sheets under Carrier Test on the main web menu.
9012	Carrier Testing as above, for two distinct mutations. \$500. When the proband has two distinct mutations both parents must be tested for both mutations.
902	Prenatal diagnosis for one or two mutations. \$2000. Includes duplicate analysis, testing for maternal cell contamination, and internal control tests. Specimens include 10 mg CVS, 10 mL AF if >15 wks, 2 confluent CV or AF culture flasks. TAT is usually 2 weeks if <u>parental</u> controls have been successfully tested in advance.

- [Click here for the test request form. Use the small box for known familial mutations on the last page of the test menu](#)
- [For specimen requirements click here to go to Amnio/CVS/Cultures under Send a Specimen](#)
- [Click here for prenatal diagnosis CPT codes and pricing](#)