



Patient Name Jane Doe	DOB	Sex Female	MRN	Invitae #
Clinical Team	Report Date	Sample Type Blood	Sample Collection Date	Sample Accession Date

Test Performed

Sequence analysis and deletion/duplication testing of the 288 genes listed in the results section below.

- Invitae Comprehensive Carrier Screen

Reason for Testing

Patient/partner is pregnant (Est. due date: 07.21.2019)

INVITAE COMPREHENSIVE CARRIER SCREEN RESULTS

About this test

This carrier test evaluated 288 genes for genetic changes (variants) that are associated with an increased risk of having a child with a genetic disorder. Knowledge of carrier status for one of these disorders may provide information that can be used to assist with family planning and/or preparation.

Result



POSITIVE

Summary

This test shows the presence of clinically significant genetic change(s) in this individual in the gene(s) indicated below. No other clinically significant changes were identified in the remaining genes evaluated with this test.

Results	Gene	Variant(s)	Inheritance	Partner testing recommended
CARRIER: Alpha-thalassemia	HBA1/ HBA2	HBA1: Deletion (Entire coding sequence)	Autosomal recessive	Yes
CARRIER: Bardet-Biedl syndrome (BBS10-related)	BBS10	c.850C>T (p.Gln284*)	Autosomal recessive	Yes
CARRIER: Gaucher disease	GBA	c.1448T>C (p.Leu483Pro)	Autosomal recessive	Yes

Next steps

- See the table above for recommendations regarding testing of this individual's reproductive partner.
- Even for genes that have a negative test result, there is always a small risk that an individual could still be a carrier. This is called "residual risk." See the table below for residual risks, which presumes a negative family history of the disorders listed.
- Genetic counseling is recommended to further explain the implications of this test result and assess family health history, which may point to health information that merits additional consideration.
- All patients, regardless of result, may wish to consider additional screening for hemoglobinopathies by complete blood count (CBC) and hemoglobin electrophoresis, if this has not already been completed.
- Individuals can register their tests at <https://www.invitae.com/patients/> to access online results, educational resources, and next steps.