Foresight®
Carrier Screen

We help you care for *every* hopeful parent

Guide *every* parent-to-be in planning for the path ahead with genetic insights

Myriad genetics

Health. Illuminated.

Every hopeful parent deserves reliable genetic insights

The goal of carrier screening is to detect couples who are at risk of passing down serious inherited conditions.

The clinical utility of carrier screening is to help at risk couples understand their risk(s) and consider their reproductive,[1] diagnostic testing, newborn screening, and treatment options.

ACOG and ACMG recommend carrier screening to ***every*** patient considering pregnancy or who is already pregnant.[1,2]

Understanding carrier screening status can help families get to a diagnosis, and possibly treatment, faster.

72% of rare diseases are genetic[3]

7.3 – number of physicians visited before getting a rare disease diagnosis[4]

4.8 years – average time it takes to diagnose a rare disease from symptom onset[4]

18% – infant hospitalizations in the US due to recessive genetic conditions[5]

Foresight® Carrier Screen helps you identify couples—of every ancestry—who are at risk to pass down serious, prevalent, clinically-actionable inherited conditions to their children, giving all your patients clear answers to guide their journey.

1 in 22

1 in 22 couples (screened with the Universal panel) identified at-risk for serious inherited conditions - the highest published detection rate[6]

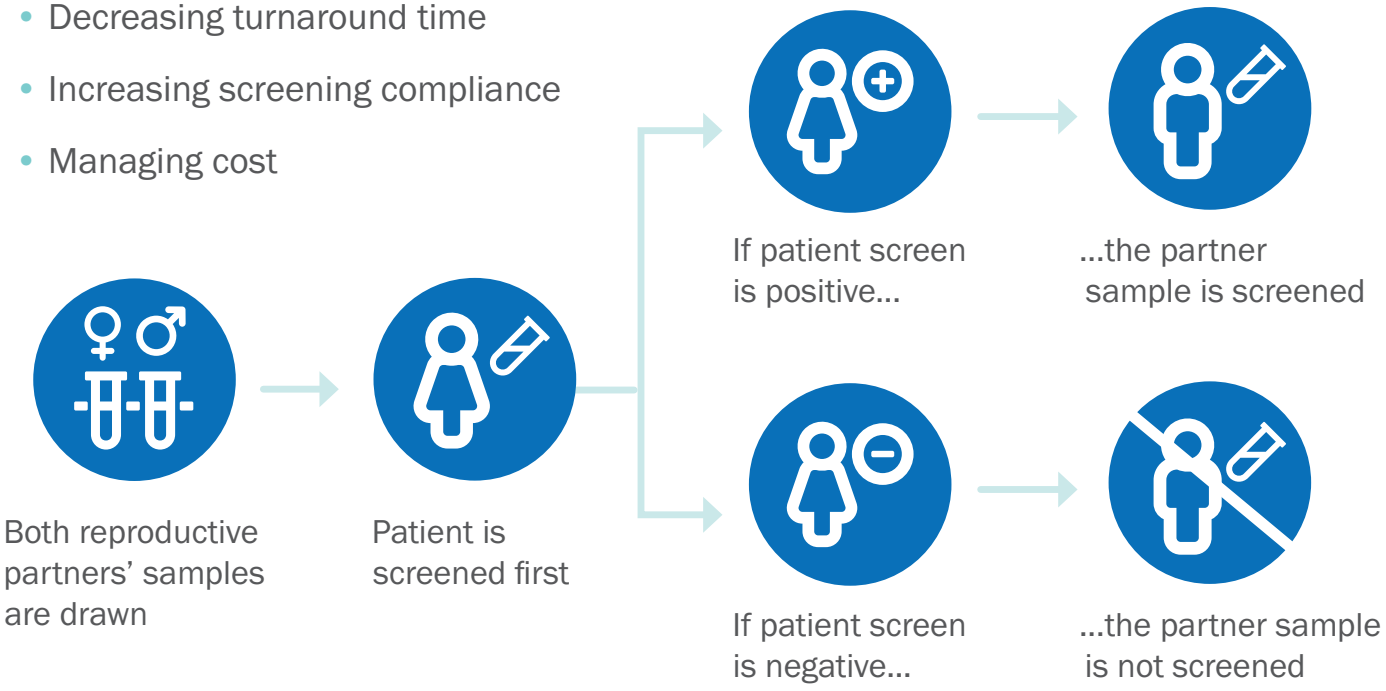
>99%

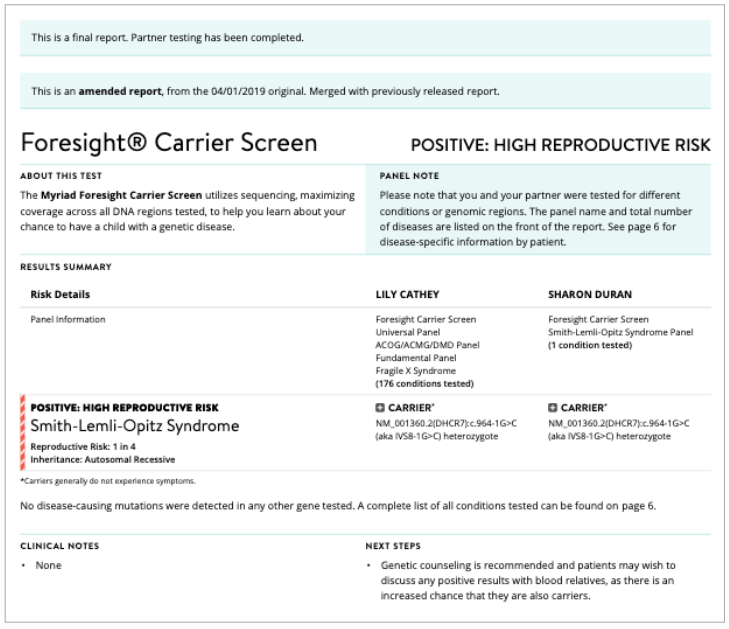
>99% detection rate for the vast majority of genes on our panels[7]

Simplify screening for *every* couple with tandem reflex and merged reports

Tandem reflex streamlines partner carrier screening by:

- Reducing follow-up coordination
- Decreasing turnaround time
- Increasing screening compliance
- Managing cost





This is a final report. Partner testing has been completed.

This is an **amended report**, from the 04/01/2019 original. Merged with previously released report.

Foresight® Carrier Screen — **POSITIVE: HIGH REPRODUCTIVE RISK**

ABOUT THIS TEST

The **Myriad Foresight Carrier Screen** utilizes sequencing, maximizing coverage across all DNA regions tested, to help you learn about your chance to have a child with a genetic disease.

PANEL NOTE

Please note that you and your partner were tested for different conditions or genomic regions. The panel name and total number of diseases are listed on the front of the report. See page 6 for disease-specific information by patient.

RESULTS SUMMARY

Risk Details	LILY CATHEY	SHARON DURAN
Panel Information	Foresight Carrier Screen Universal Panel ACOG/ACMG/DMD Panel Fundamental Panel Fragile X Syndrome **(176 conditions tested)**	Foresight Carrier Screen Smith-Lemli-Opitz Syndrome Panel **(1 condition tested)**
POSITIVE: HIGH REPRODUCTIVE RISK Smith-Lemli-Opitz Syndrome **Reproductive Risk: 1 in 4** **Inheritance: Autosomal Recessive**	**CARRIER*** NM_001360.2(DHCR7):c.964-1G>C (aka IVS8-1G>C) heterozygote	**CARRIER*** NM_001360.2(DHCR7):c.964-1G>C (aka IVS8-1G>C) heterozygote

*Carriers generally do not experience symptoms.

No disease-causing mutations were detected in any other gene tested. A complete list of all conditions tested can be found on page 6.

CLINICAL NOTES

- None

NEXT STEPS

- Genetic counseling is recommended and patients may wish to discuss any positive results with blood relatives, as there is an increased chance that they are also carriers.

Merged reports offer a clear view of overall reproductive risk

You and your patients have access to our Board-Certified Genetic Counselors

Carrier screening panels designed with *every* patient in mind

Myriad Genetics pioneered the first expanded carrier screening to maximize detection of at-risk couples. Our goal is to produce not simply more, but meaningful clinical information. We offer three standard panels:

Fundamental

Cystic fibrosis and spinal muscular atrophy

Fundamental Plus

Guidelines-focused set of 14 genes

Universal

176 genes associated with serious and prevalent inherited conditions

Panels may be customized based on clinical needs

Meet the Pantier family

Real-world impact:

Ashley Pantier shares the positive health impact the Foresight® Carrier Screen has had on her family

SCAN ME

1. American College of Obstetricians and Gynecologists' Committee on Practice Bulletins—Obstetrics; Committee on Genetics; Society for Maternal-Fetal Medicine. Screening for Fetal Chromosomal Abnormalities: ACOG Practice Bulletin, Number 226. Obstet Gynecol. 2020;136(4):e48-e69. doi:10.1097/AOG.0000000000004084 **2.** Gregg, A.R., Aarabi, M., Klugman, S. et al. Screening for autosomal recessive and X-linked conditions during pregnancy and preconception: a practice resource of the American College of Medical Genetics and Genomics (ACMG). Genet Med 23, 1793–1806 (2021). https://doi.org/10.1038/s41436-021-01203-z **3.** Nguengang Wakap, S., Lambert, D.M., Olry, A. et al. Estimating cumulative point prevalence of rare diseases: analysis of the Orphanet database. Eur J Hum Genet 28, 165–173 (2020). https://doi.org/10.1038/s41431-019-0508-0 **4.** Global Genes, Accurate Diagnosis of Rare Diseases Remains Difficult Despite Strong Physician Interest, Published March 6, 2013, Accessed March 21, 2022 https://globalgenes.org/2014/03/06/accurate-diagnosis-of-rare-diseases-remains-difficult-despite-strong-physician-interest/ **5.** Kingsmore S. Comprehensive carrier screening and molecular diagnostic testing for recessive childhood diseases. PLoS Curr. 2012;4:e4f9877ab8ffa9. Published 2012 May 2. doi:10.1371/4f9877ab8ffa9 **6.** Hogan GJ, Vysotskaia VS, Beauchamp KA, et al. Validation of an Expanded Carrier Screen that Optimizes Sensitivity via Full-Exon Sequencing and Panel-wide Copy Number Variant Identification. Clin Chem. 2018;64(7):1063-1073. doi:10.1373/clinchem.2018.286823 **7.** Based on internal data

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