preparent® exon select

carrier test

progenity®

A focused, right-sized carrier test that's optimized for detection.

We designed a new carrier test that includes many of the most commonly inherited disorders. This high clinical utility test is focused on detection and actionability.

Curation guided by ACOG 690

- Disorders were selected based on recommended clinical inclusion criteria offered by ACOG Committee Opinion 690.[1]

Maximize identification of positive carriers

- This test includes many of the most common hereditary disorders found in the general population.

High actionability for better outcomes

- Test for disorders that cause serious health problems in infancy or childhood, and which benefit from early diagnosis and treatment.

Identify more carriers for more prepared patients.

Fast Facts

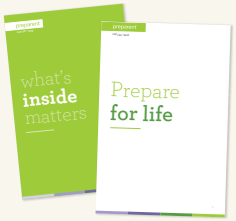
Test 13 genes with average carrier frequency >1 in 60[2]	Up to 23% positivity rate[3]
1 tube of blood or buccal swab	Results in 14-17 days

Clinical sensitivities of 99% for most disorders across all ethnicities.[4]

Patient support tools

Patient Education Displays

Patient Education Video



Patient Brochures

Carrier testing helps everyone prepare for early intervention when needed, which can lead to better health outcomes. Many of these disorders are included on newborn screening panels because of the serious health issues they can cause if not diagnosed and treated early.

Disorder	Gene	TRIO	Carrier frequency (general population)	May be included in newborn screening panels[5]	Better outcomes with early detection & treatment	Medication & treatment plan
Cystic fibrosis	*CFTR*	•	1 in 25	✓	✓	
Spinal muscular atrophy	*SMN1*	•	1 in 54	✓	✓	
Fragile X syndrome	*FMR1*	•	1 in 250		✓	
Hemoglobinopathies			1 in 20	✓	✓	
Alpha-thalessemia	*HBA1/HBA2*					
Beta-hemoglobinopathies including sickle cell anemia	*HBB*					
Congenital disorder of glycosylation type IA	*PMM2*		1 in 124			
Galactosemia	*GALT*		1 in 110	✓	✓	
Gaucher disease	*GBA*		1 in 153	✓	✓	
Glycogen storage disease type IA	*G6PC*		1 in 177		✓	
Medium-chain acyl-CoA dehydrogenase deficiency	*ACADM*		1 in 35	✓	✓	
Phenylalanine hydroxylase deficiency	*PAH*		1 in 65	✓	✓	
Smith-Lemli-Opitz syndrome	*DHCR7*		1 in 100		✓	
Tay-Sachs disease	*HEXA*		1 in 250			

LEGEND medication · dietary treatment · specialized therapy · symptom management

1. Carrier screening in the age of Genomic Medicine. Committee Opinion No. 690. American College of Obstetrics and Gynecology. Obstet. Gynecol. 2017.
2. Fragile X is not included in average calculation due to X-linked inheritance. Disease incidence is equivalent to other disorders on the list. Not tested in males.
3. Progenity, data on file.
4. Baylor Genetics, data on file.
5. Newborn screening panels vary from state to state.

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