

Screening for **7,535 mutations** related to **18 Common** **Recessive Disorders**

Maternal carrier screening combines Next-generation sequencing (NGS) with GAP-PCR to detect both point mutations and deletion variants, including HBA1 and HBA2 gene deletions.

The 18 conditions are selected based on three key criteria:

Most common diseases

Carrier frequency $\geq 1/200$

Severe diseases

Causing cognitive or physical impairment, or have an onset early in life

Actionable diseases

Requiring surgical or medical intervention, antenatal intervention to improve perinatal outcomes

18 Recessive Disorders based on American College of Medical Genetics and Genomics (ACMG) guideline⁽³⁾

"All pregnant patients and those planning a pregnancy should be offered Tier 3 carrier screening for autosomal recessive (carrier frequency $\geq 1/200$) and X-linked conditions."

Performance ^{(1),(2)}

95-99%

Sensitivity

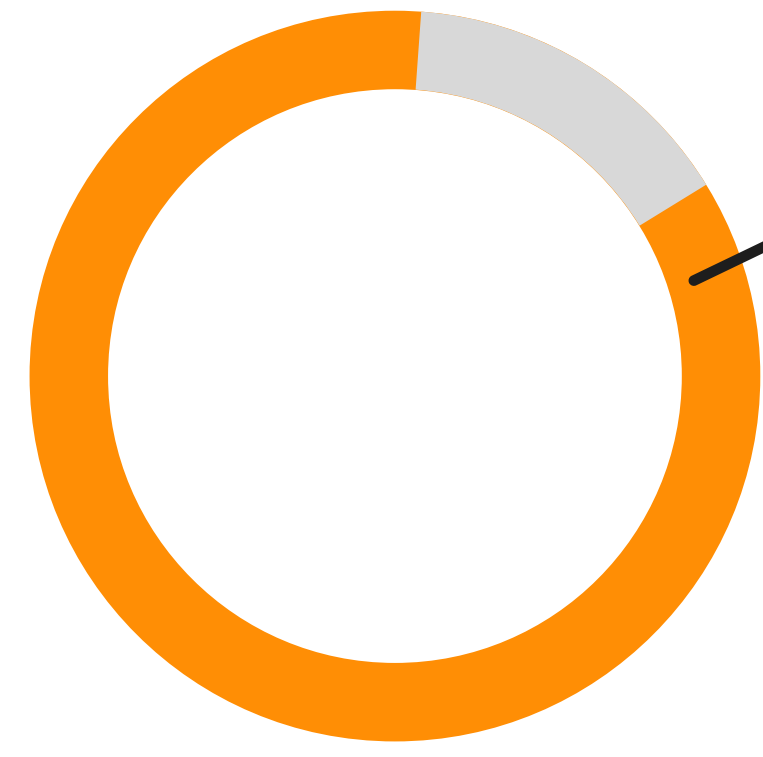
>99%

Specificity

• Scope of The Test

Disease	Gene	Clinical Burden	Treatment
Alpha-Thalassemia	HBA1, HBA2	- Minor to severe phenotype: hydrops fetalis, stillbirth, severe anemia. - Women with HbH disease require high-risk perinatal care. - Women who are carrying a fetus with Hb Bart's hydrops fetalis are at risk for potentially serious complications during the pregnancy.	Lifelong blood transfusion, splenectomy, Iron overload treatment.
Beta-Thalassemia	HBB	Minor to severe phenotype: Severe anemia exhibit within the first two years of life, jaundice, failure to thrive.	Lifelong blood transfusion, splenectomy, Iron overload treatment.
Citrin deficiency	SLC25A13	- Low birth weight, intrahepatic cholestasis, failure to thrive in neonates and children. - Adult-onset recurring episodes of hyperammonemia and associated neuropsychiatric symptoms.	Supplement diet with fat-soluble vitamins and use of lactose-free and medium-chain triglyceride (MCT)-enriched formula.
G6PD deficiency	G6PD – X-linked	Acute hemolytic anemia (AHA), which may be life-threatening especially in children.	Preventative measures.
5 alpha reductase deficiency	SRD5A2	Newborns are genetically male but are assigned female at birth based on their external genitalia.	Surgical repair hormonal therapy.
Pompe disease	GAA	Within the first three months of life: rapidly progressive muscle weakness, diminished muscle tone, respiratory deficiency, hypertrophic cardiomyopathy, failure to thrive.	Enzyme Replacement Therapy, Supportive Therapies.
Phenylketonuria	PAH	Severe intellectual deficit and neuropsychiatric complications.	Low-protein diet and use of a Phe-free medical formula as soon as possible after birth.
Galactosemia	GALT	Early diagnosis and treatment with a lactose-restricted (dairy-free) diet is absolutely essential to avoid profound intellectual disability, liver failure and death in the newborn period.	Lactose-restricted (dairy-free) diet Breastfeeding is contraindicated.
Wilson disease	ATP7B	Cirrhosis, neurological disorders, premature death.	Life-long medication to lower copper levels.
Pendred syndrome	SLC26A4	The main clinical manifestations are sensorineural deafness, goiter, and congenital hypothyroidism. The degree of hearing loss is variable: it can be mild to moderate, and severe to profound in other patients.	Adjunctive treatment: hearing aids, cochlear implants, thyroid function therapy.
Primary carnitine deficiency	SLC22A5	Metabolic disorders of the carnitine cycle and carnitine transport are typically characterized by early-onset cardiomyopathy in infancy, often accompanied by weakness and low muscle tone, developmental delay, and recurrent hypoglycemic seizures and/or coma.	Oral carnitine supplementation for life.
Niemann-Pick disease	SMPD1	Hepatosplenomegaly, usually detected by three months of age; over time, the liver and spleen become enlarged. Growth retardation, respiratory failure, and early death before age 3.	Symptomatic treatment
Cystic fibrosis	CFTR	Multisystem disease affects the epithelium of the respiratory tract, exocrine pancreas, intestine, hepatobiliary system, and exocrine sweat glands. Pulmonary disease is the major cause of death.	Symptomatic treatment
Charcot-Marie-Tooth disease, axonal, type 2S	IGHMBP2	Infants often have a history of intrauterine growth retardation, low birth weight, weak cry, weak sucking, and failure to thrive, with inspiratory stridor recurrent episodes of dyspnea or apnea, cyanosis, and loss of deep tendon reflexes. Kyphosis/scoliosis, foot deformities, and joint contractures are common features.	Symptomatic treatment
Polycystic kidney disease	PKHD1	The clinical spectrum includes varying degrees of renal failure, mild to life threatening neonatal respiratory distress due to pulmonary hypoplasia, hyponatremia, hypertension, and susceptibility to urinary tract infections.	Symptomatic treatment
Hemophilia A	F8 – X-linked	Bleeding abnormalities begin to appear when affected male infants begin to walk. However, infants with hemophilia are at risk of intracranial or extracranial hemorrhage and other bleeding complications.	Factor VIII supplementation. Treatment may be given after a bleeding episode or prophylactically, to prevent bleeding.
Adrenoleukodystrophy	ABCD1 – X-linked	The disease is characterized by endocrine dysfunction (hypoadrenalism/testicular failure), progressive myelopathy, peripheral neuropathy, and progressive leukodystrophy. It affects both males and females.	Adrenal insufficiency is treated with hydrocortisone. Leukodystrophy can be treated with allogeneic hematopoietic cell transplantation (HCT) in the early stages.
Ornithine transcarbamylase	OTC – X-linked	Male infants are usually normal at birth but within a few days they may develop poor feeding, low muscle tone, and lethargy, which rapidly progresses to drowsiness and coma. Seizures and hyperventilation may also occur. If left untreated, severe encephalopathy develops with a high risk of death.	Lifelong protein restriction and nitrogen-removing drugs, liver transplant.

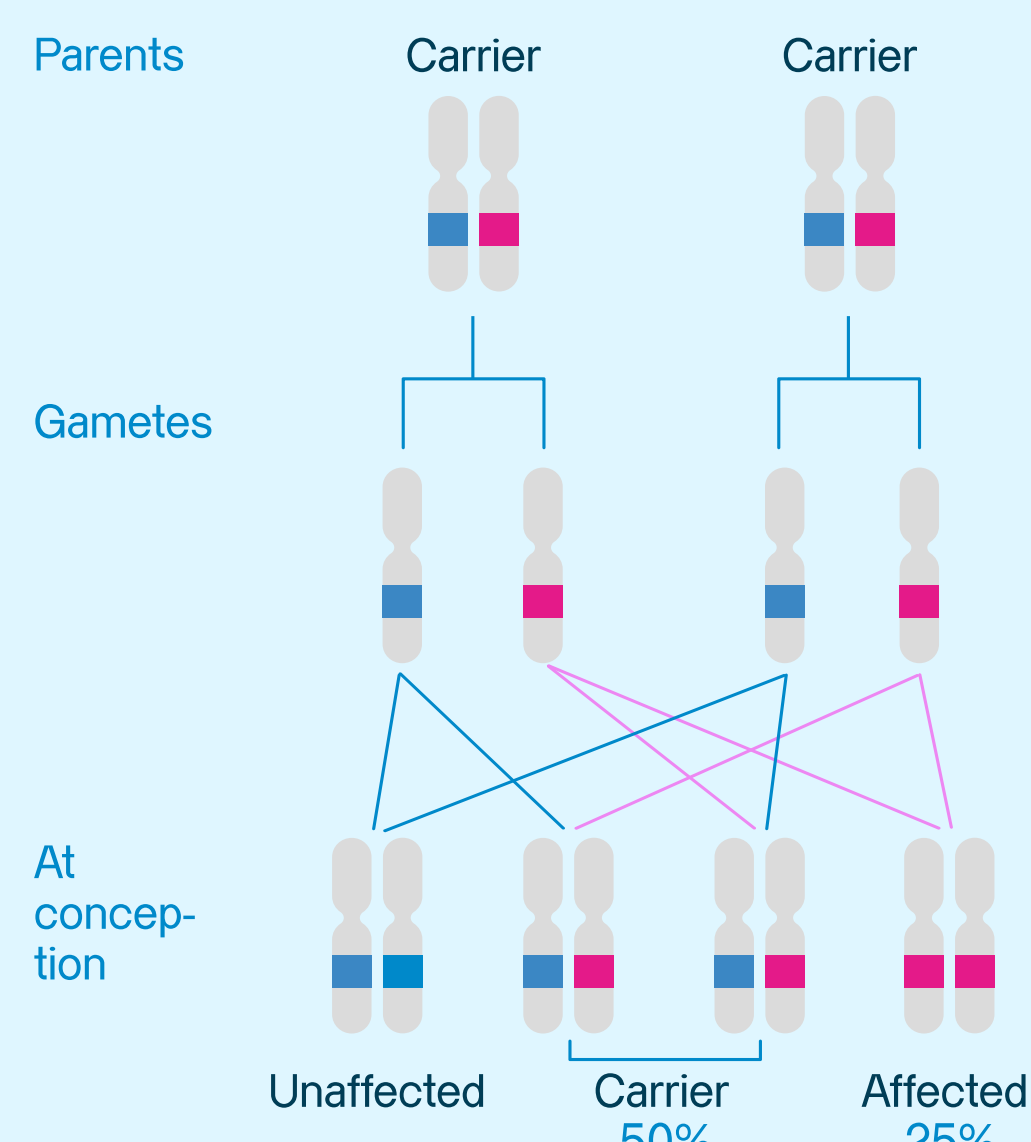
Recessive Inheritance



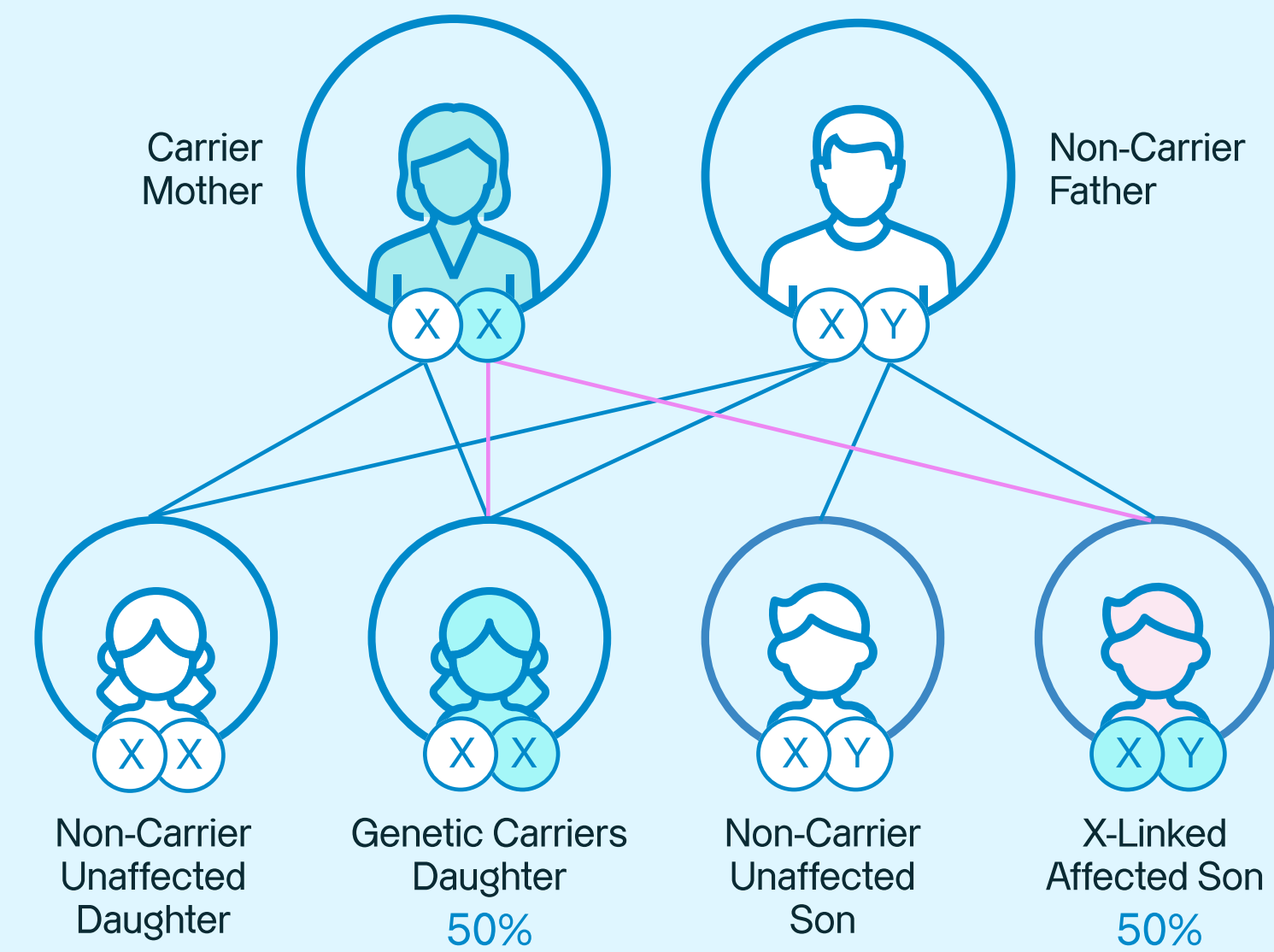
More than **80%** of children with genetic disorders are born to healthy parents or parents with no family history of the disorder.*

*Blythe SA, et al. Clin Biochem. 1984;17(5):277–283.

Autosomal recessive disease inheritance



X-linked recessive disease inheritance



Clinical utilities

Pregnancy management plan for severe diseases

- Severe forms of alpha thalassemia: Hb Bart's syndrome (+)
- Women diagnosed with HbH disease and women who are carrying a fetus with Hb Bart's hydrops fetalis are at risk for potentially serious complications during the pregnancy, require high-risk perinatal care

Early intervention and treatment immediately after birth

- Inform the doctor at birth about the positive screening result of parents
- Perform genetic testing diagnoses children immediately after birth
- Examination and consultation with the neonatal pediatrician

Preventive plan for each pregnancy

- Prenatal/postnatal diagnosis
- IVF - PGT- M (Preimplantation Genetic Testing for Monogenic disorders)

Cascade screening for family (parents, siblings, children)

- Risk of transmitting disease-causing genes to future generations

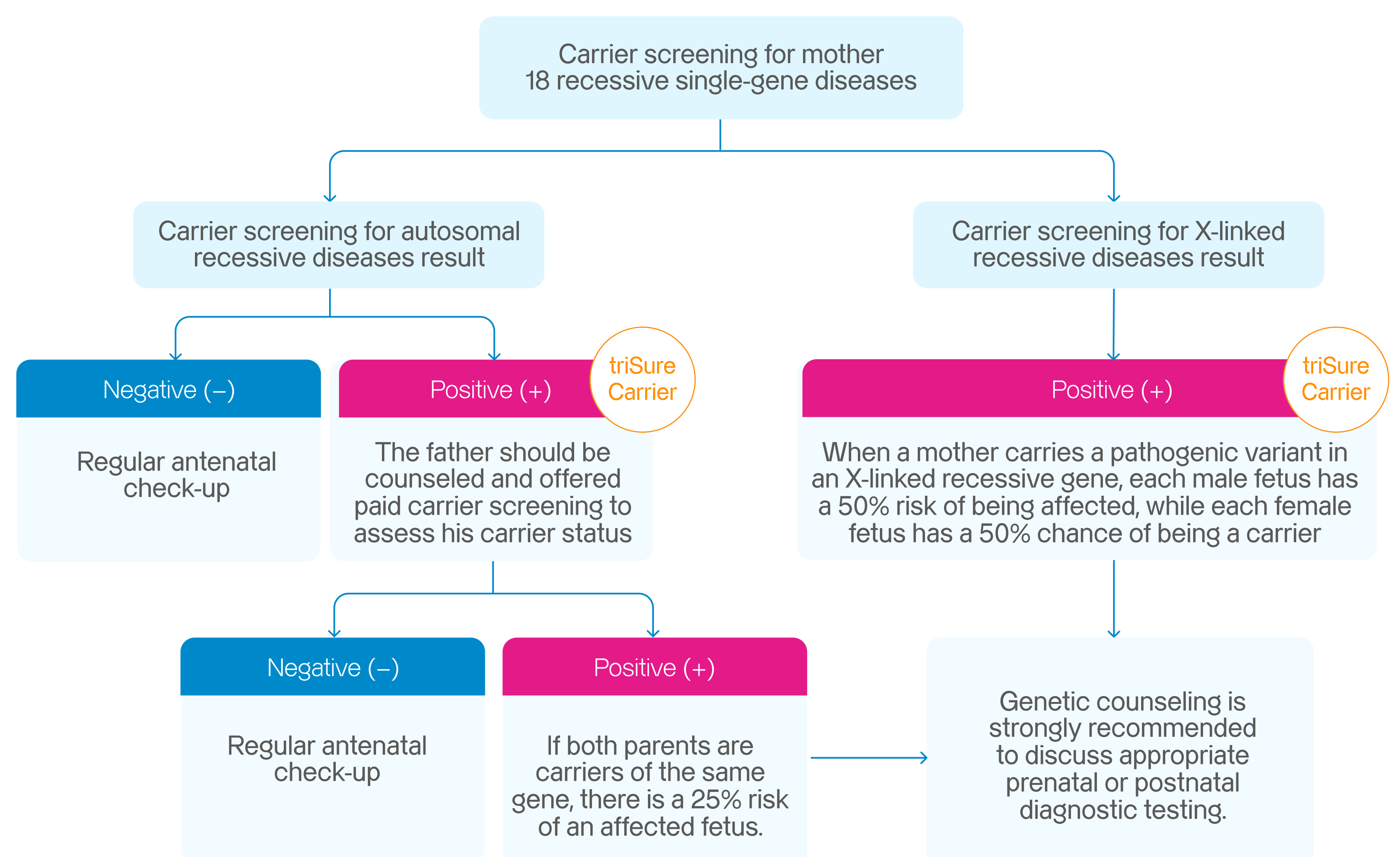
Testing process

- Blood sample collection
- NGS + GAP-PCR sequencing at Gene Solutions laboratory
- Test Results (TAT-8 days**)

**TAT may vary depending on the country.

As of 2025, Gene Solutions has delivered over **700,000 maternal recessive carrier reports**, empowering healthcare providers with actionable genetic data to enhance prenatal and pediatric care outcomes.

Results & Follow-up Recommendations



1. Doan, P.-L., et al. "Detection of maternal carriers of common α -thalassemia deletions from cell-free DNA." Scientific Reports. 2022;12:13581.
 2. Tran, N. H., et al. "Genetic landscape of recessive diseases in the Vietnamese population from large-scale clinical exome sequencing." Human Mutation. 2021;42:1229–1238.
 3. Gregg, A. R., 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)." Genetics in Medicine. 2021;23:1793–1806.

For more information or to request a brochure, please contact us:
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