GENETIC CARRIER SCREENING

Genexplore Diagnostics & Research Centre Pvt. Ltd.

"Exploring the spiral of life"

is a test to check if individuals carry genetic mutations that could be passed on to their children, potentially causing inherited disorders

Basic

Male
- Karyotyping
- CFTR
- Y-Chromosome micro deletion
- Sperm DNA fragmentation

Female
- Karyotyping
- CFTR
- DMD
- Fragile-X

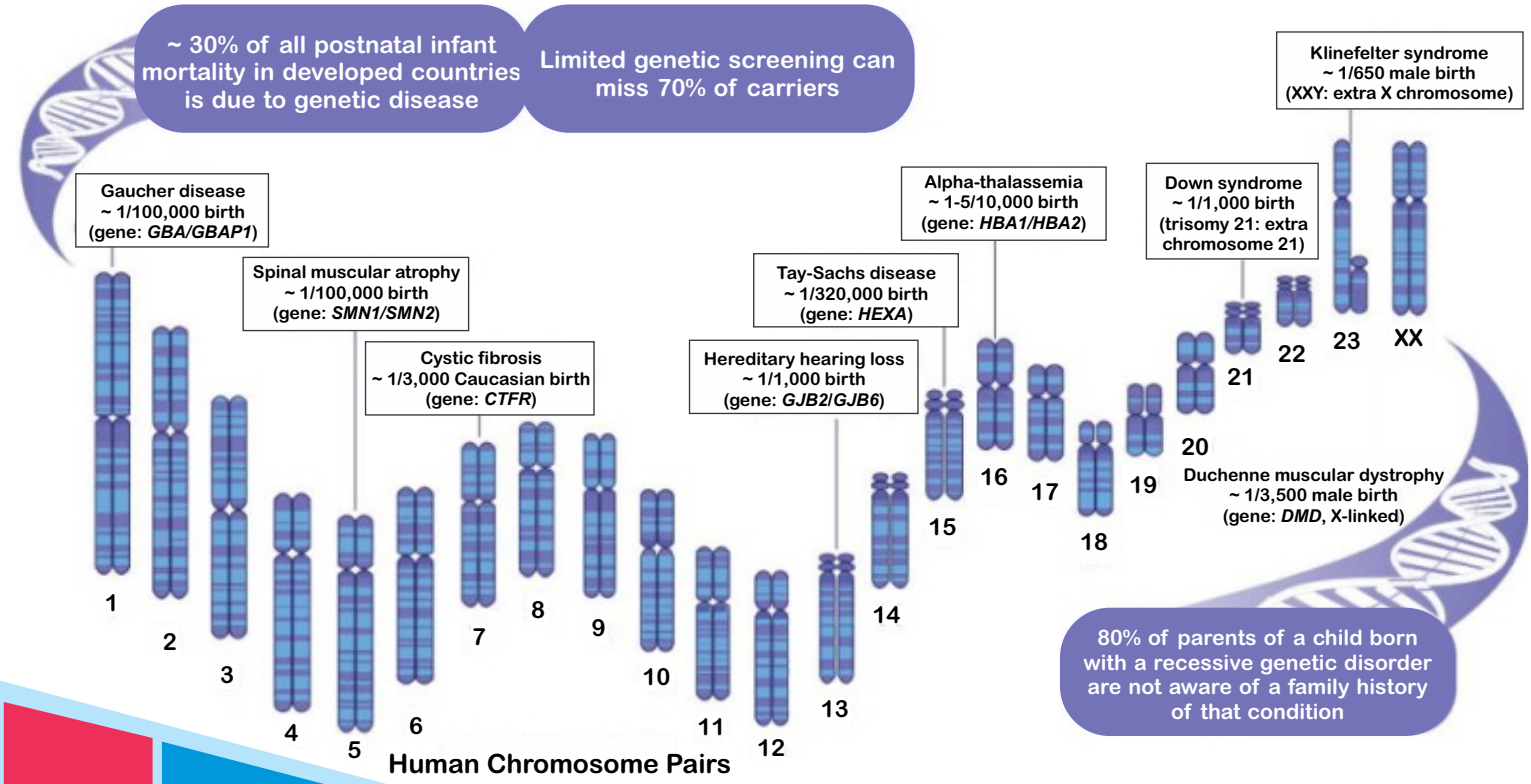
Advanced

Male & Female
- Karyotyping
- Expanded carrier screening (DMD, SMA, α-Thalassemia and β-Thalassemia)

Genetic Carrier Screening include:

~ 30% of all postnatal infant mortality in developed countries is due to genetic disease

Limited genetic screening can miss 70% of carriers

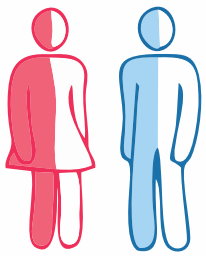


80% of parents of a child born with a recessive genetic disorder are not aware of a family history of that condition

14,000 Amplicons	418 Inherited Disorders	
420 Genes	33 genes with single exon-level resolution	• American College of Obstetricians and Gynecologists (ACOG) recommended panel.
	>36000 SNVs, Indels and CNVs	• Complete automated workflow reduces human errors. • Validated Carrier Reporter Software for robust analysis.

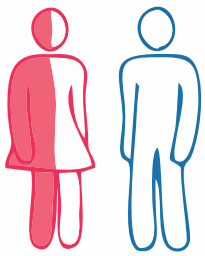
Autosomal recessive condition:

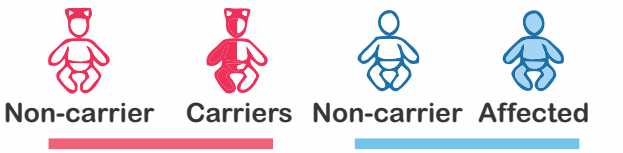
When female and male both are carriers

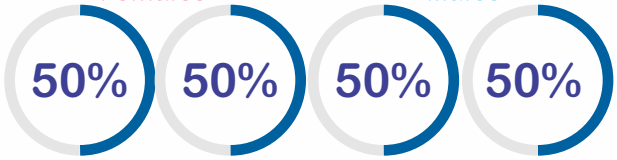


X-linked recessive condition:

When female is a carrier

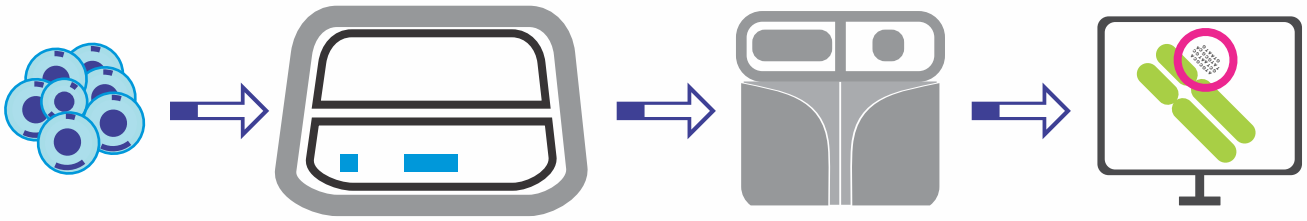






- If you are born to parents who both carry the same autosomal recessive gene, you have a 25% (1 in 4) chance of inheriting the abnormal gene from both parents and developing the disease. You have a 50% (1 in 2) chance of inheriting one abnormal gene. This would make you a carrier.
- In X-linked recessive condition, if mother has the mutated X-linked gene then daughters are usually not affected and are called carriers.
- Son will be affected if they inherit the mutated X-linked gene from their mother.
- Fathers can not pass X-linked recessive conditions to their sons

Workflow:



Sample required : EDTA Blood

Test method : NGS Based

TAT : 15 days

Genexplore™

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