Neuberg DIAGNOSTICS

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Neu INSIGHTS

Neuberg DIAGNOSTICS CENTER FOR GENOMIC MEDICINE

LUMOUS CARRIER SCREENING

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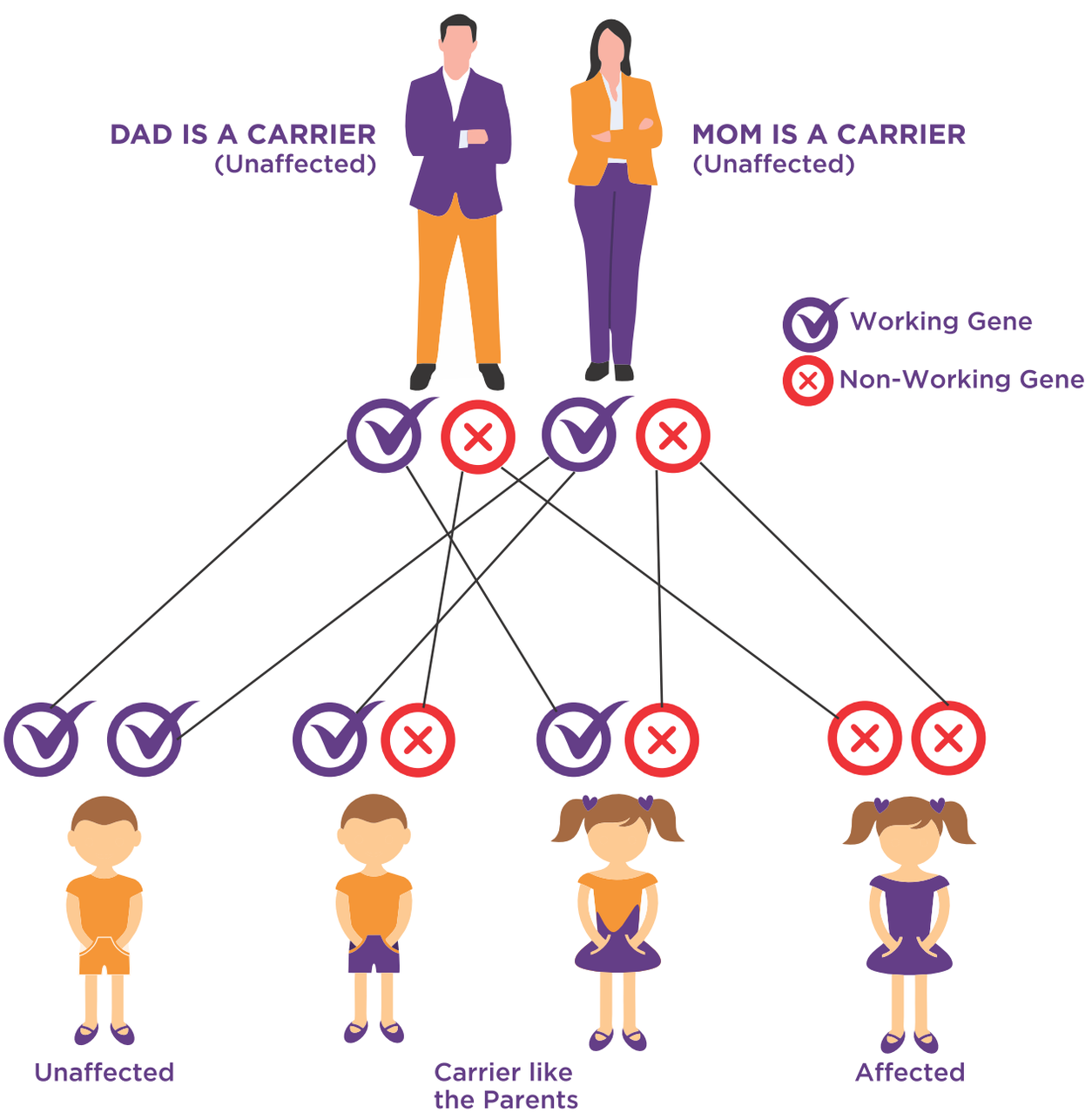
What is Carrier Screening?

Genetic Carrier Screening can provide valuable information for the genetic health of a family.

- Molecular testing is used to identify individuals and couples at risk for a child with an Autosomal or X-linked Recessive genetic disorders. It helps to provide risks associated with severe genetic conditions in the family.
- When both the husband and wife are carriers for the same condition, there is a risk (25% chance) of having an affected child with every pregnancy.

Who are Carriers?

Carriers are asymptomatic individuals who may carry a genetic variant that can increase the risk of having a child with a genetic disorder. Any individual can be a carrier of a genetic condition.



Who should consider carrier screening?

Carrier screening can be offered to couples who:

01 **Have a family history of a genetic disorder**

02 **Are themselves having a genetic disorder**

03 **Bad obstetric history (IUD/ neonatal deaths / >2 children with "suspected" genetic condition)**

04 **Are consanguineously married**

05 **Are part of a community at high risk of a genetic disease**

06 **Are considering egg/sperm donation for IVF**

Why to do Carrier Screening?

It provides the couple with knowledge regarding their pregnancy so that they can make informed choices regarding reproductive options and further management

What disorders should be screened?[1]

- Common Autosomal and X-linked recessive disorders as recommended by consensus groups based on population / ethnicity-based studies.
- It can also include disorders based on strong family history.

The options provided by LUMOUS

LUMOUS provides flexibility in the amount of information needed.

Tests/Methodology	Lumos Focus		Lumos Comprehensive		Lumos Plus	
	Female	Male	Female	Male	Female	Male
Sequencing of >2500 genes [NGS] **	✓	✓	✓	✓	✓	✓
Spinal Muscular Artophy (SMA) [MLPA]	✓	✓	✓	✓	✓	✓
Congenital Adrenal Hyperplasia (CAH) [MLPA & Sequencing]			✓	✓	✓	✓
Duchenne Muscular Dystrophy (DMD) [MLPA]	✓		✓		✓	
Fragile X [TP-PCR]	✓		✓		✓	
Alpha Thalassemia [MLPA]					✓	✓
Hemophilia A (including F8 Intron 1/22 Inversion)					✓	

** Genes implicated in >2900 OMIM based Autosomal and X-linked recessive disorders.

Why LUMOUS?

Comprehensive analysis of all OMIM defined recessive disorders. (>2,900 conditions)

Superior technology providing >95% coverage of all included genes.

Insightful analysis and reporting based on phenotype when family history is positive.

LUMOUS with a purpose!

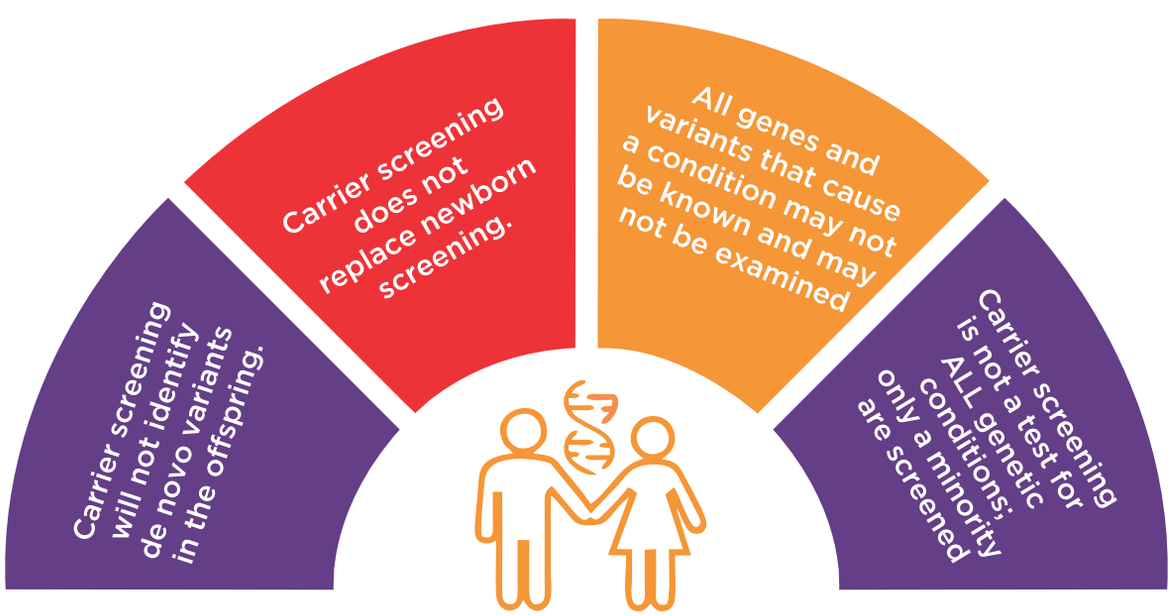
IF Positive
Carrier status in the couple is confirmed & can predict risk of recurrence.

IF Negative
Reduced chance of having an affected child but does not eliminate the risk.

No confusion/No difficult choices.

- Only disease-causing variants (pathogenic or likely pathogenic) are reported.
- Does not report variants of uncertain significance unless indicated by significant family history.

No test is perfect!



(***Please contact the lab to inquire for the gene/disorder list covered under NGS)

How can the information from the carrier screening be used?

- All LUMOUS tests are offered with FREE genetic counseling services.
- A genetic counselor can discuss the results with the family and provide further information about :
  1. The conditions which were screened positive.
  2. The risk for future pregnancies.
  3. Choice of reproductive options for parents.

LUMOUS process flow

Pre -Test Genetic Counselling | **Sample Collection** | **Lumous Processing** | **Report Release** | **Post -Test Genetic Counselling**

References:

1. Gregg AR, Aarabi M, Klugman S, Leach NT, Bashford MT, Goldwaser T, Chen E, Sparks TN, Reddi HV, Rajkovic A, Dungan JS; ACMG Professional Practice and Guidelines Committee. 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. 2021 Oct;23(10):1793-1806.
2. Stevenson, R. E. (Ed.). (2015). Human malformations and related anomalies (No. 66). Oxford University Press.
3. Taber, K. A. J., Beauchamp, K. A., Lazarin, G. A., Muzzey, D., Arjunan, A., & Goldberg, J. D. (2019). Clinical utility of expanded carrier screening: results-guided actionability and outcomes. Genetics in Medicine, 21(5), 1041-1048.
4. Westemeyer, M., Saucier, J., Wallace, J., Prins, S. A., Shetty, A., Malhotra, M., ... & Billings, P. (2020). Clinical experience with carrier screening in a general population: support for a comprehensive pan-ethnic approach. Genetics in Medicine, 22(8), 1320-1328.

Our Services

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Reproductive Genetics

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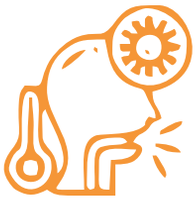

Infectious Disorders

Pharmacogenomics

Research Services

PARTNERS IN HEALTH

DR. SHEETAL SHARDA

Director - Clinical Genomics
Development & Implementation
M.D. Pediatrics
D.M. Medical Genetics
sheetal.sharda@ncgmglobal.com

DR. UDHAYA KOTECHA

Head of Division Inherited Disorders (NGS)
Clinical Geneticist
M.D. Pediatrics
Fellowship in Medical Genetics
udhaya.kotecha@ncgmglobal.com

DR. PRIYA RANGANATH

Consultant Medical Geneticist
MS (ObG), DrNB (Medical Genetics)
priya.ranganath@ncgmglobal.com

DR. MEHUL MISTRI

Scientist- Inherited Genetics & Metabolism
PhD Biochemistry
mehul.mistri@ncgmglobal.com

DR. SANDIP SHAH

Consultant Pathologist
M.D (Pathology & Bacteriology)
Laboratory Director
drsandip@neubergdiagnostics.com

FOR MORE DETAILS, CONTACT US AT

079 61618111
079 40408181

ncgmglobal.com

Neuberg DIAGNOSTICS

● India ● UAE ● South Africa ● USA