CGT Carrier Genetic Test

Igenomix® WITH SCIENCE ON YOUR SIDE

OUR NEW UNIVERSAL APPROACH TO EXPANDED CARRIER SCREENING (CS) USING WHOLE EXOME SEQUENCING

	CGT Bank	CGT Plus	CGT Exome
Methodology	**WHOLE EXOME SEQUENCING (WES)**		
Type of panel	**Exclusive Panel for Gamete Donors** ACOG	**Expanded Panel**	**Premium Expanded Panel**
Genes	**M: 7 genes** **F: 71 genes** (include 64 X-linked)	**M: 455 genes** **F: 519 genes** (include 64 X-linked)	**M: 1,979 genes** **F: 2,043 genes** (include 64 X-linked)
Variants	~3,800	~20,000	>50,000
Numbers of diseases	Up to 75	Up to 570	More than 2,200
Estimated carrier rate (%)*	~11%	~55%	~67%
Estimated mean of mutations/individual**	1	1.7	2.7
Mean depth	150X	150X	150X
Complementary tests	**M/F:** CYP21A2, HBA1/2, SMN1 **F only:** DMD, FMR1, F8	**M/F:** CYP21A2, HBA1/2, SMN1 **F only:** DMD, FMR1, F8	**M/F:** CYP21A2, HBA1/2, SMN1 **F only:** DMD, FMR1, F8
Sample	**Blood**	**Blood**	**Blood**
TAT	**20 working days**	**20 working days**	**20 working days**

* In-house data base of 30,000 tests
**Estimated mean of positive individuals

M: male; F: Female

Why choose our CS Exome based?

CLINICAL ADVANTAGE

- Allows for testing of **All known recessive conditions.**
- Increases the overall detection rate minimizing the global residual risk.

MATCHING

- Maximizes IVF applications, **matching possible with ALL genetic lab tests in the market.**
- **Simplifies** competitor CS panel mirroring as no resequencing is required to provide matching information.

UPGRADES

- **Any upgrade possible** at a later date if required.

REANALYSIS

- **Exome Sequencing offers added value for future analysis of a given patient.**
- Provides analytical possibilities in an adverse event of a newborn with a genetic condition.

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CGT is an advanced genetic test performed before pregnancy that determines the risk of having a child with a genetic disease.

It helps prevent disorders without cure.

2-5% of couples

carry a mutation in the same gene

Ropers, 2012. Martin J et al. 2015
May vary among populations

1,000 live births

10 Affected

Estimated global prevalence

According to the World Health Organization (WHO)

20%

infant mortality in developed countries

Hoyert DL et al. 2000
Kingsmore S. 2012

88%

Of children born with a genetic disease have no family history for the conditions.

Archibald et al; 2018

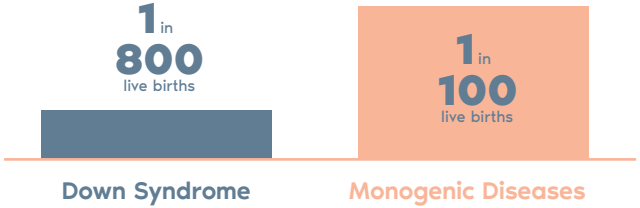
The American College of Obstetricians and Gynecologists (ACOG) makes the following recommendations:

ACOG
The American College of Obstetricians and Gynecologists

Information about genetic carrier screening should be provided to every pregnant woman.

THE MOST COMMON MONOGENIC DISORDERS DETECTED WITH THE CGT TEST ARE:	PROPORTION OF CARRIERS
Cystic fibrosis	1 in 25
Spinal Muscular atrophy	1 in 50
Autosomal recessive polycystic kidney disease	1 in 70

GLOBAL PREVALENCE IN LIVE BIRTHS OF DOWN SYNDROME VS MONOGENIC DISEASES



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