CGT Carrier Genetic Test

Igenomix® WITH SCIENCE ON YOUR SIDE

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

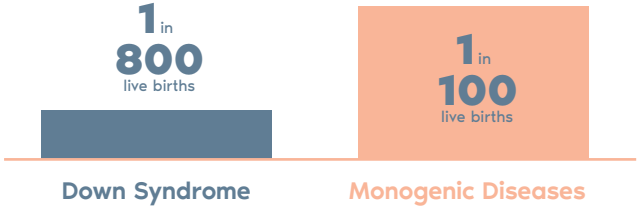
The American College of Obstetricians and Gynecologists (ACOG) makes the following recommendations:

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



www.igenomix.com

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There is a different CGT test for every patient's needs

	CGT Essential	CGT Plus	CGT Exome
Methodology	**MULTIGENE PANEL NGS**		**WES**
Features	Cost-effective solution	Based on the recommendation of medical societies ACOG The American College of Obstetricians and Gynecologists	Whole exome compatible with most carrier platforms in the market
Genes	19	306	1590
Variants	2236	16592	30592
Numbers of diseases	20	352	>1600
Estimated carrier rate (%)*	18%	54.8%	62.7%
Estimated mean of mutations/individual**	1.08	1.46	2.28
Mean depth	1000X	350X	100X
Complementary tests	FMR1, SMN1	HBA, F8, FMR1, SMN1	HBA, F8, FMR1, SMN1, DMD, CYP21A2
Sample	Blood or saliva		
TAT	14 working days	20 working days	20 working days

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