

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 	Expanded Panel	Premium Expanded Panel
 Genes	M: 7 genes F: 73 genes (include 66 X-linked)	M: 470 genes F: 536 genes (include 66 X-linked)	M: 1,989 genes F: 2,055 genes (include 66 X-linked)
 Variants	~3,800	>30,000	>50,000
 Numbers of diseases	Up to 75	More than 500	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 or saliva	Blood or saliva	Blood or saliva
 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.

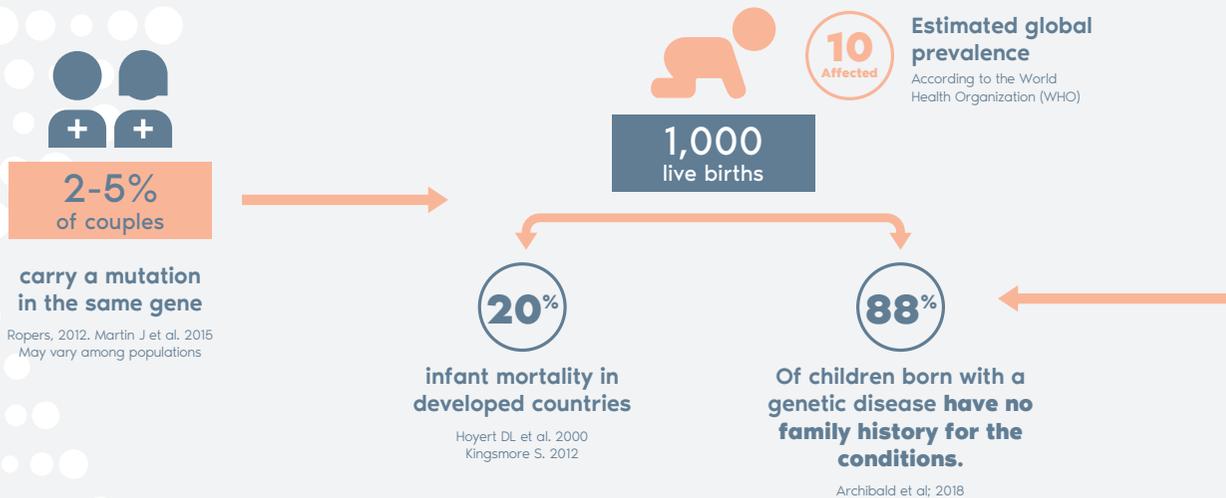


REANALYSIS

- **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.

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.



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

