

Our new universal approach to expanded carrier screening (CS) using Clinical Exome Sequencing (CES)

	CGT Bank	CGT Plus	CGT Exome	CGT Mirror*	CGT One/Sync*
Methodology	Clinical Exome Sequencing (CES)			Clinical Exome or Whole Exome Sequencing	
Panel	Exclusive Panel for Gamete Donors	Expanded	Premium Expanded	Custom	Matching against Known Carriers
TAT (Business Days)	15	15	15	15	15
Reported Genes	Customised 65 XL genes	463 AR 65 XL	1648 AR 65 XL	Custom	AR genes, Custom (Sync include 65 XL)
Reported Diseases	Custom	Up to 570	>2,000	Case Dependent	Case Dependent
Estimated Carrier Rate (approx. %)	11% (Basic)**	55%	67%	Case Dependent	Case Dependent
Mean Depth	150X	150X	150X	150X	150X
Complementary tests (to NGS)	AR: FXN; CYP c.955C>T XL: FMR1; F8 (in22 inv)	AR: FXN; CYP c.955C>T XL: FMR1; F8 (in22 inv)	AR: FXN; CYP c.955C>T XL: FMR1; F8 (in22 inv)	If required	If required
Sample Type	Blood/Saliva	Blood/Saliva	Blood/Saliva	Blood/Saliva	Blood/Saliva

* Estimated that <5% of CGT Sync requests will require WES instead of CES; for CGT Mirror, this will vary depending on panel

** Genes CFTR, SMN1, HBB, HBA1/2, GJB2, CYP21A2 – general EU population
AR: autosomal recessive; XL: X-linked

Why choose the carrier screening test from Igenomix?



CLINICAL ADVANTAGE

- Allows for testing of **frequently prevalent recessive conditions**.
- Increases the overall detection rate minimizing the global residual risk.
- Upgraded CGT now delivers Copy Number Variations (CNV) analysis to detect relevant gene deletions and duplications for increased diagnostic sensitivity.



MATCHING

- Maximizes IVF applications, **matching possible with ALL genetic lab tests in the market**.
- Simplifies** competitor CS panel mirroring.



UPGRADES

- Upgrades are possible** at a later date if required.

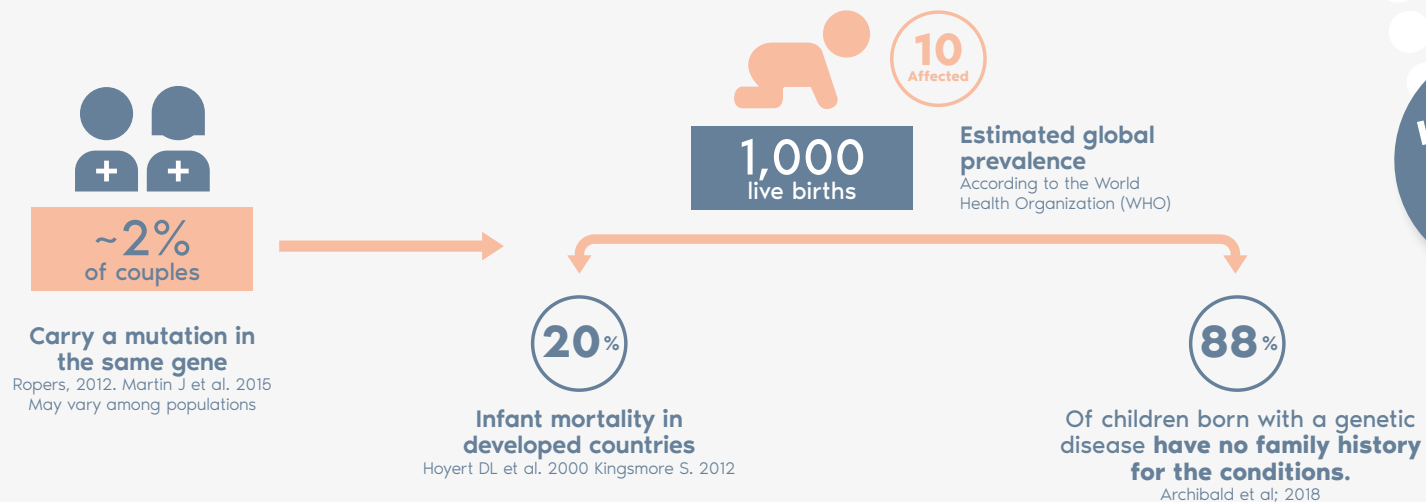


REANALYSIS

- 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.

COMPLIANCE

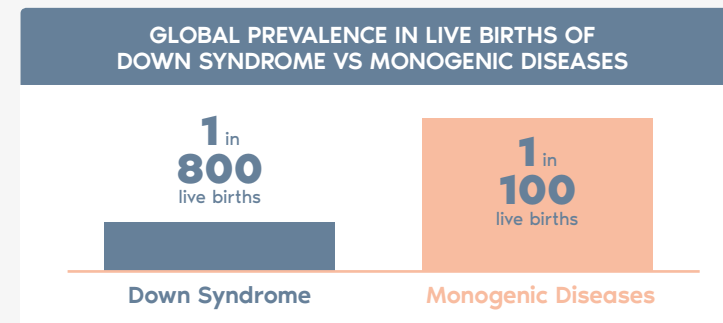
Upgraded CGT testing using CES for clinically focused carrier screening and preconception risk detection



www.igenomix.co.uk

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



(* Carriers identified in the general Caucasian population.)