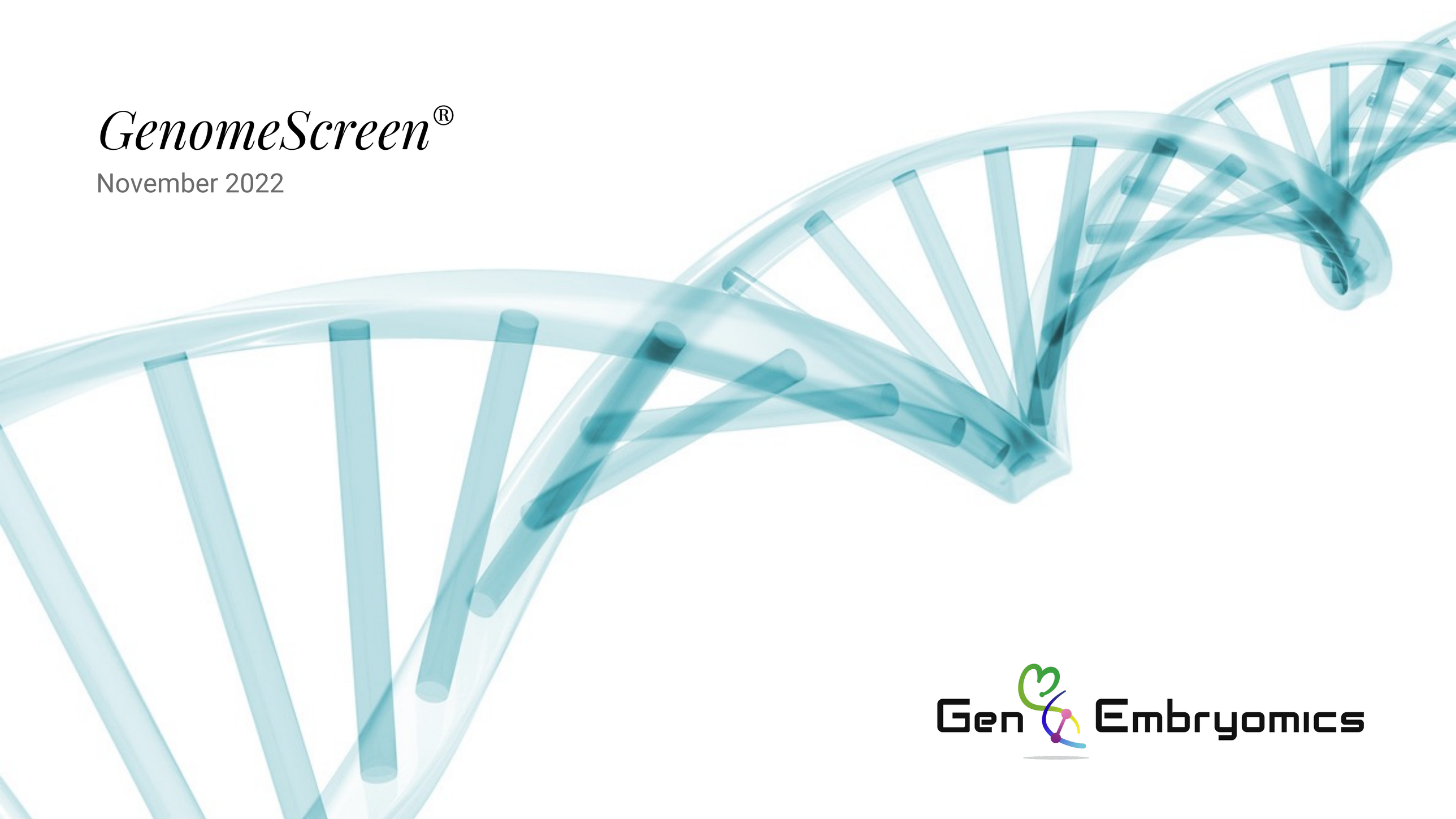
GenomeScreen®

November 2022



Gen Embryomics

About Us

We have developed the first *de novo* mutation screening test for IVF embryos. This is pre-implementation genome screening for whole genome sequencing (PGT-WGS).

GenomeScreen® is a comprehensive test that screens trophectoderm biopsied IVF embryos for every genetic disease.

Executive Team

NICK MURPHY | PHD

GenEmbryomics CEO/Founder

Co-inventor of the COVID-19 Risk test and Polygenic Risk Multi-Test (Genetic Technologies). First to directly sequence the human HLA system from a single chromosome.

SANTIAGO MUNNÉ | PHD

Executive Director & Senior Advisor

PhD in Human Genetics (University of Pittsburgh). CIO at Overture Life. Founder of: Reprogenetics, Recombine, Phosphorus, MedAnswers, Overture Life, G1 Sciences. Advisory Board (currently): Overture Life, Phosphorus, MedAnswers, Butterfly & Sama.

MONICA PETICA | MSC

Chief Clinical Officer

Clinical leader, quality management & curation.

Monash IVF, Victorian Clinical Genetics Services (VCGS), Australian Forensic Police Services (Victoria, South Australia, Northern Territory)

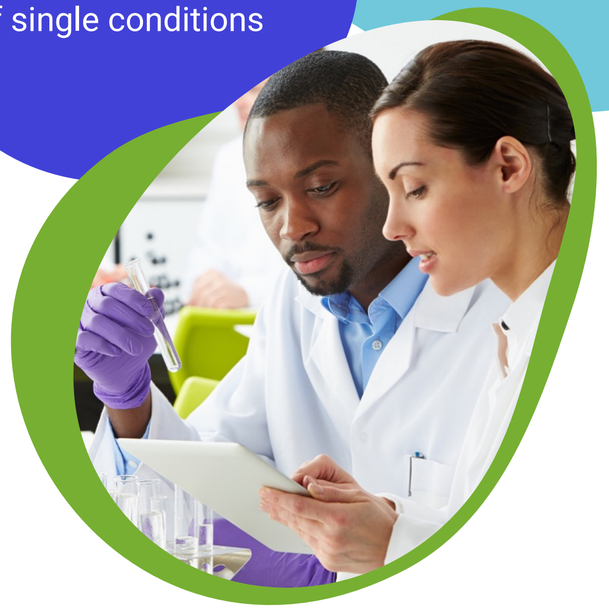
The Problem

De novo mutations spontaneously occurring at fertilization accounts for over **4,525 genetic diseases that are routinely missed by standard PGT.** These diseases destroy innumerable lives and lead to **massive costs to the healthcare system.**

300,000
IVF cycles in USA alone

4500+
Genetic conditions which could occur

50%
Patients pay for tests of single conditions



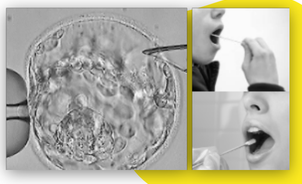
Solution

GENOMESCREEN®

- World first to screen for 4500+ genetic conditions at the same cost of doing a single test.
- The 50% of IVF clients already paying for screening of 1 genetic condition can pay the same amount for 4500+ conditions.

Test Overview

DNA COLLECTED



IVF Clinic collects the DNA samples from parent donors and the embryo trophectoderm cells and sends the samples to the lab

DNA SEQUENCED

Lab provides whole genome sequencing for the DNA samples and uploads the FASTQ files for each sample to the GenEmbryomics HIPAA compliant cloud server

CLOUD BIOINFORMATICS

1. Mapping sequences into a BAM file
2. Short variant calling
3. CNV calling
4. Repeat Expansion calling

VARIANT CURATION

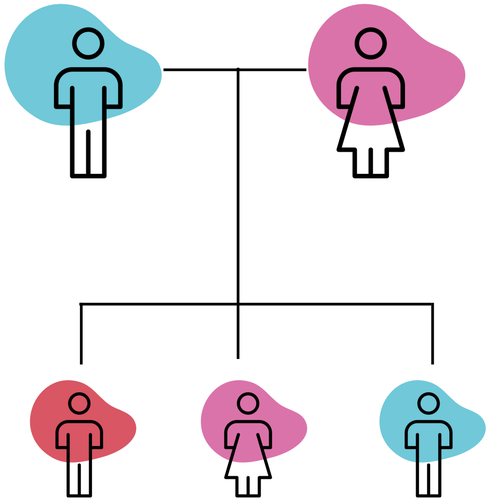
The data is curated by a variant curator and reported with a clinical geneticist.

CLINICAL REPORT

The test report is sent to IVF clinic for IVF doctor to inform patients about the genetic health of the embryo

GenomeScreen®

- The genomes of both parents are required for categorising each variant.
- Filtering is performed for false-positive base misincorporations and sequencing artefacts.
- Variants are “annotated” from >50 annotation sources, pathogenicity prediction algorithms and ACMG guidelines.
- Embryo transfer/screening recommendations provided based on severity and age of onset for fatal, disabling and early onset (objective) and contextual (subjective) pathogenicity conditions where patients may need ethics approval for screening.



Affected child = *De novo* mutation/AR disorder/CNV/mosaic/sex-linked

ALL CLASSES OF PATHOGENIC MUTATION

Spontaneous (de novo) mutations

Specificity	99.99%
Accuracy	99.99995760%
Sensitivity	>80% *de novos* detected compared to 0% with other tests
NPV	99.99999703%
False Positive Rate	0.39%
False Negative Rate	0.65%

Transmitted mutations (>99.9999%)

SNV’s, Indels, MNVs

Copy number variants

Aneuploidies & translocations & triploidy

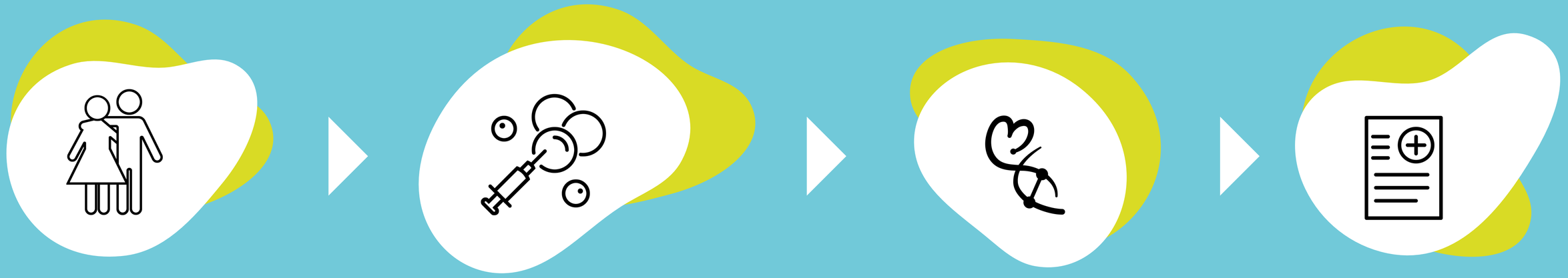
Trinucleotide repeats (e.g. FRM1)

Mosaicism (high accuracy)

Translocations

Polygenic risk

Our Process



1 Couples seeking IVF treatment

2 IVF Clinic

3 GenomeScreen® Report by GenEmbryomics

4 Clinical Report sent to IVF doctor

Embryo biopsy & parent DNA Samples collected & sent to the lab for whole genome sequencing

- Clinical recommendation for embryo
- Pathogenic variants affecting embryo
- Embryo recommendation

50%

Patients pay for tests of single conditions

PANACEA™

GenomeScreen®

For collecting and screening DNA embryo before implantation

Couplet ™

GenomeScreen®

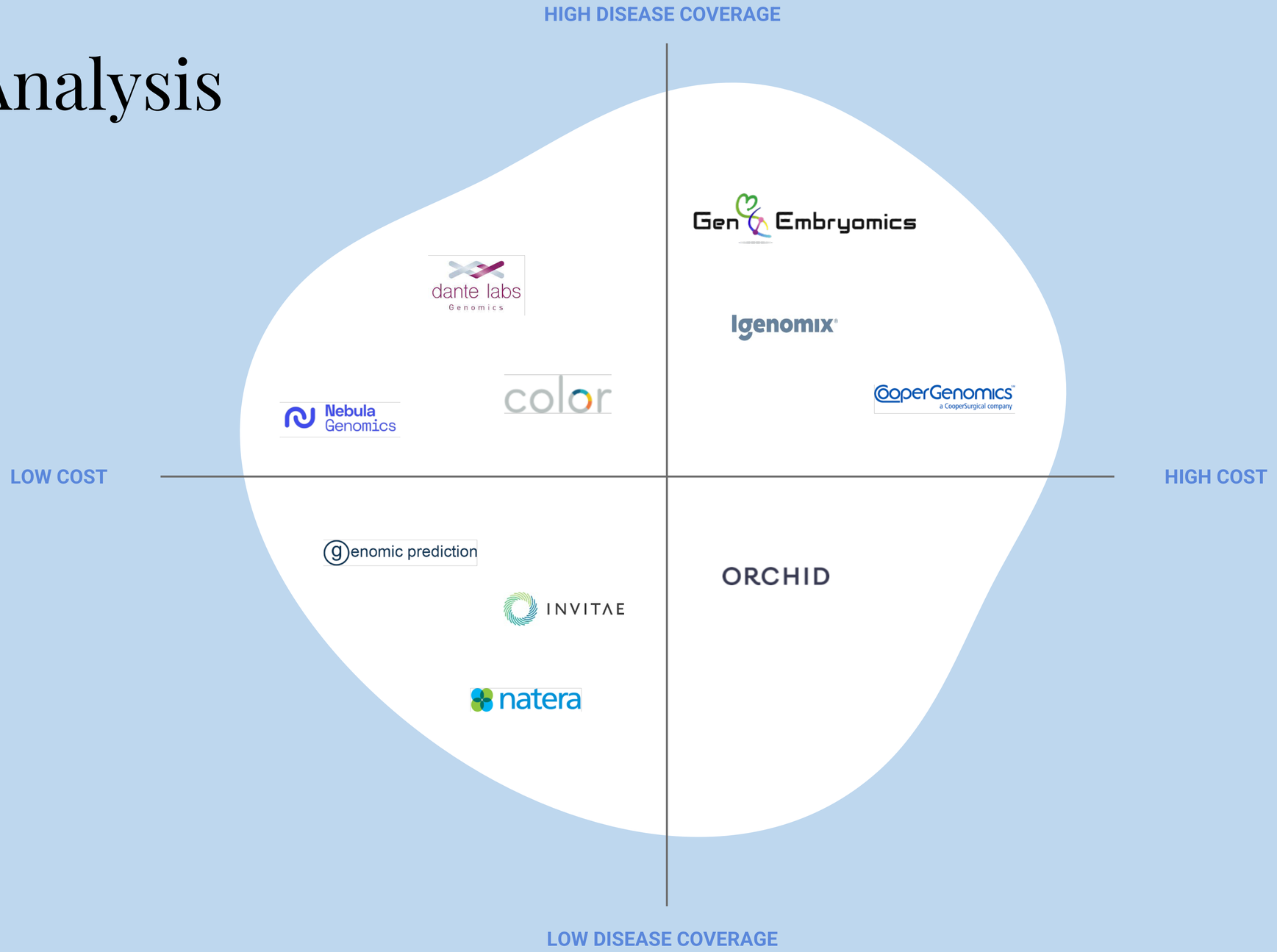
For collecting and screening DNA of couples

GenomeScreen Ova™

GenomeScreen®

For collecting and screening DNA of IVF Oocytes

Competitor Analysis

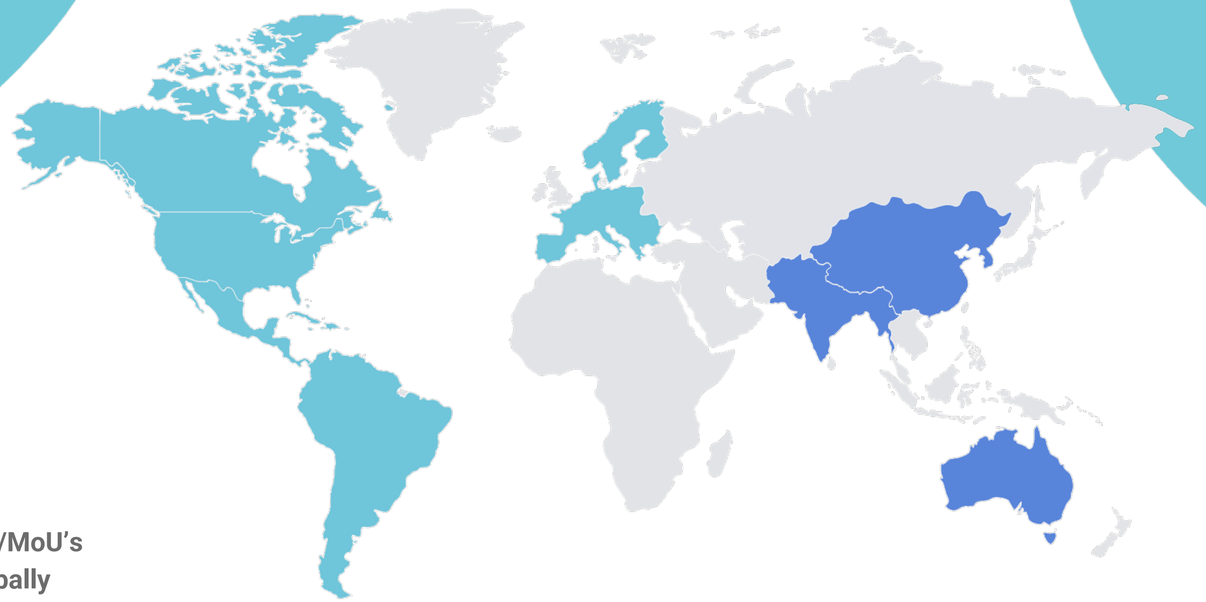


GenomeScreen® vs. Competitors

	GenEmbryomics	genomic prediction	CooperSurgical	natera	Igenomix	ORCHID
4,525 genetic diseases/test	✓	✗	✗	✗	✗	✗
Single mutations	✓	✓	✓	✓	✓	✓
Chromosome number aneuploidy	✓	✓	✓	✓	✓	✓

GenomeScreen® is the only provider of PGT-WGS

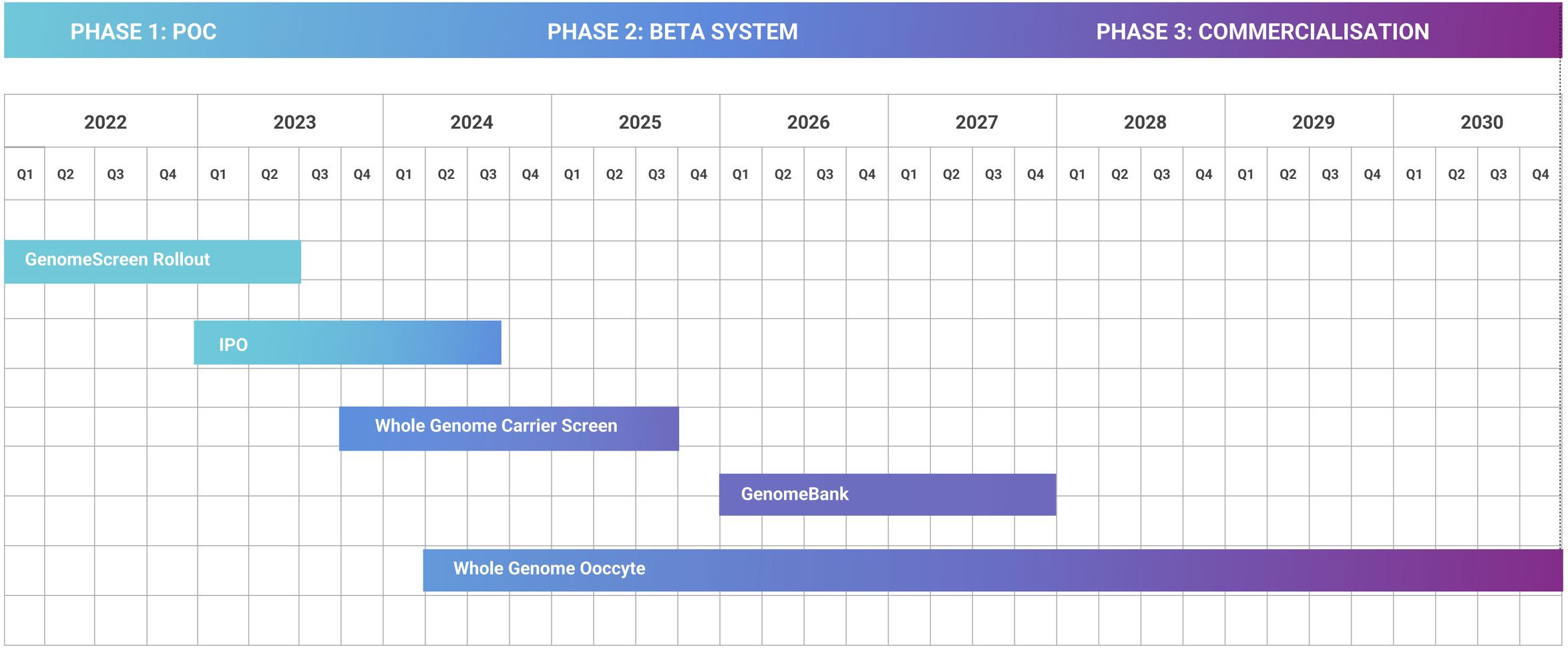
Global Clinical Partners



10 LOI/MoU's globally

75 IVFs Clinics Globally

Appendix C - Roadmap For Product Development



Connect with us!

contact-us@genembryomics.com

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