

Genetic Testing 101

Outline

1. Understanding the terms.
 - a. Pre-2019
 - i. Preimplantation Genetic Testing (PGT)
 1. Preimplantation Genetic Screening (PGS)
 2. Preimplantation Genetic Diagnosis (PGD).
 - b. Current Terminology
 - i. PGS is now Preimplantation Genetic Testing for Aneuploidies (PGT-A).
 - ii. PGD is now
 1. Preimplantation Genetic Testing for Monogenic/Single-Gene Disorders (PGT-M)
 2. Preimplantation Genetic Testing for Chromosomal Structural Rearrangements (PGT-SR)
2. PGT-A
 - a. What is PGT-A?
 - b. Aneuploidy can cause what adverse outcomes?
 - c. How many chromosomes can be screen for?
 - d. Who should consider having this testing?
 - e. What can PGT-A show us?
 - i. Whole Chromosomal that are extra or missing.
 - ii. Difficult to detect a balanced translocation.
 - iii. What is a Polyploidy?
 - iv. Segmental aneuploidy? Smaller deletions or duplications
 1. Because the missing or extra piece of chromosome is less severe than a whole extra or missing chromosome, segmental aneuploidies have a higher chance or resulting in a child born with physical or intellectual disabilities.
 - f. What do we mean by the resolution of the testing?
 - g. Is it possible to determine from which parent (the egg or sperm) the chromosome abnormality originated? Why might this be important?
 - h. What are the different testing platforms that can be used with PGT-A?
 - i. **N**ext **G**eneration **S**equencing (**NGS**)
 - ii. **S**ingle **N**ucleotide **P**olymorphisms (**SNPs**)
 - i. Mosaicism
 - i. What is mosaicism? (the presence of a mixture of chromosomally normal and abnormal cells)
 - ii. Not related to maternal age. Occurs after fertilization.
 - iii. Are there different degrees of mosaicism?
 - iv. Does the presence of mosaicism impact the implantation rate, miscarriage rate, and birth defect rate?
 - j. What does the research show on how PGT-A impacts the success rate of IVF?

- k. How is the test performed?
 - l. How accurate is PGT-A testing? What is the risk of false negatives and false positives?
3. Preimplantation Genetic Testing for Monogenic/Single-Gene Disorders (PGT-M)
- a. What is PGT-M? (looks for specific inherited conditions controlled by a single gene)
 - b. Alternative options
 - c. Who should consider having this testing?
 - d. Does PGT-M impact the success rate of IVF?
 - e. Does this testing process differ from PGT-A?
 - f. How accurate is PGT-M testing?
4. Preimplantation Genetic Testing for Chromosomal Structural Rearrangements (PGT-SR)
- a. What is PGT-SR?
 - b. Who should consider having this testing?
 - c. Does this testing process differ from PGT-A or PGT-M?
 - d. How accurate is PGT-SR testing? Same range mid to high 90%
5. Important Additional Information
- a. Patients and health care providers should be aware that a “normal” or negative preimplantation genetic test result is not a guarantee of a newborn without genetic abnormalities.
 - b. Importance of genetic counseling.