Results Summary

- **NORMAL EMBRYOS** 1, 3
- **ABNORMAL EMBRYOS** 2

Results Interpretation

<table>
<thead>
<tr>
<th>EMBRYO #</th>
<th>KIT #</th>
<th>BIOPSY DATE</th>
<th>CHROMOSOME RESULT*</th>
<th>INTERPRETATION*</th>
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</thead>
<tbody>
<tr>
<td>1</td>
<td>025400</td>
<td>08/01/2018</td>
<td>Normal</td>
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<td>025400</td>
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<td>3</td>
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<td>Normal</td>
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</tbody>
</table>

**The results provided are based on the assessment of loss or gain of chromosomal material. This assay does not assess balanced changes within or between chromosomes. Please see Comments for details of assay resolution.**
Definitions

Abnormal (aneuploid): An abnormal number of chromosomes was detected.

Complex Abnormal: A complex abnormal result indicates there are five or more abnormal findings. The presence of multiple chromosome abnormalities is not expected to be compatible with life.

Haploid: Only one set of chromosomes (23) is present as opposed to the typical two sets (46) in a normal sample. This type of chromosome abnormality is not compatible with life.

Indeterminate: This interpretation indicates that a reliable result cannot be achieved with the sample submitted, as the data quality did not meet the proper quality thresholds. In the case of an indeterminate sample, a re-biopsy is recommended if testing is still desired.

Isochromosome: An abnormal result where one arm of the chromosome is lost and the other arm is duplicated. Depending on if the centromere is duplicated, isochromosome can be abbreviated as “i” or “idic”. This is not a normal chromosome result and is not recommended for transfer.

Monosomy: One chromosome of a normal pair is absent. The report will show a “-” before the missing chromosome. The majority of monosomies are not compatible with life.

Mosaic: An abnormal result indicating that cells within the same biopsy sample have a mixed chromosome makeup. It can involve either whole chromosome or partial chromosome segment(s). Upon physician request, the report will show a “(mos)” after the chromosome result, indicating that the reported changes are mosaic. The clinical impact of a mosaic embryo varies; however, this is not a normal chromosome result and is not recommended for transfer.

No Results: This interpretation indicates that a reliable result cannot be achieved with the sample submitted. This could be due to the lack of DNA (limited material or absence of sample) or poor data quality (no intact nucleus, poor embryo quality, degraded DNA). A re-biopsy is recommended if testing is still desired.

Normal (euploid): A normal number of chromosomes (46) was detected. A normal result does not guarantee the birth of a healthy baby or a specific embryo sex.

Partial Deletion/Duplication (partial aneuploidy): A partial deletion or duplication indicates that a piece of a chromosome is missing or extra. Partial deletions and duplications may cause abnormalities in a fetus or live birth and are generally associated with physical or cognitive abnormalities.

Special considerations: This interpretation indicates that no abnormalities were detected. However, the data quality did not meet the validated quality thresholds so the accuracy of the prediction is lower than typically quoted and a re-biopsy may yield a different result.

Triploid: An extra set of chromosomes is present, so there are 3 copies of each chromosome (69) as opposed to the typical two copies (46) in a normal sample. This type of chromosome abnormality is not compatible with life.

Trisomy: The presence of an extra copy of a chromosome was detected. The report will show a “+” before the extra chromosome. The majority of trisomies are not compatible with life.

Tetraploid: Two extra set of chromosomes are present, so there are 4 copies of each chromosome (92) as opposed to the typical two copies (46) in a normal sample. This type of chromosome abnormality is not compatible with life.

Uniparental isodisomy (UPiD): Indicates that two copies of an identical chromosome were inherited from one parent and no copies from the other parent. The report will show a “UPiD” before the affected chromosome. Consequences of UPiD may include known syndromes and recessive disorders. However, not every chromosome showing UPiD has been associated with clinical consequences.

Comments

This assay is designed to assess chromosome gain or loss (aneuploidy). Preimplantation Genetic Testing (PGT) cannot detect all potential birth defects and does not guarantee a particular pregnancy outcome or embryo sex. There is a 3-5% risk in the general population of birth defects. These may occur for genetic and/or non-genetic reasons.

PGT for aneuploidy does not detect all genetic and chromosomal disorders such as those caused by single gene mutations (e.g. cystic fibrosis or sickle cell disease), all forms of uniparental disomy, all instances of mosaicism, all forms of polyploidy (loss/gain of whole sets of chromosomes), and/or structural chromosome rearrangements including balanced translocations and partial loss or gain of chromosome material (partial aneuploidy) below the resolution of this assay. This assay may be uninformative in some patients with recurrent pregnancy loss, consanguinity, and known or unknown chromosomal rearrangements below the resolution of the assay.
PGT is not 100% sensitive and specific and is intended for screening to identify the majority of embryos with a significant gain or loss of chromosomal material. False positive or negative errors may occur for reasons including but not limited to: sample mix-up or misidentification; technical problems; interfering substances; gain or loss of chromosomal material beyond the detection limit of this assay; sample contamination; the use of non-validated sample collection and handling procedures; and mosaicism (if the cells analyzed are not representative of the inner cell mass of the embryo, or there is a gain or loss of chromosome material that is not present in all cells of the biopsy sample). Validation studies showed that this assay achieves an accuracy of > 97%, specificity of > 97% and analytic sensitivity of > 95% for the detection of whole chromosome gains or losses and for detection of partial chromosome gains or losses that are ≥ 10Mb in size. Uniparental isodisomy assessment is not available for chromosomes 17, 19-22. The forms of polyploidy that are detectable include haploidy/complete UPiD, triploidy, and some forms of tetraploidy. While this assay is highly accurate, results may be impacted by artifacts present in the embryo biopsy samples.

Because PGT is a screening modality, follow up prenatal diagnosis using either chorionic villus sampling at 10-12 weeks or amniocentesis at 15-18 weeks should be considered to verify the chromosome status of an ongoing pregnancy.

Methods

PGT testing was performed in our San Francisco, CA facility located at 1400 16th Street, San Francisco, CA 94103. Samples were analyzed using a modified FAST-SeqS next generation sequencing method and associated bioinformatics pipeline validated for accurate detection of whole chromosome number, segmental (≥ 10 Mb) aneuploidy, polyploidy and UPiD (chromosomes 1-16, 18, and X).

References


Disclaimer

This test was developed and performance characteristics were determined by Invitae Corporation. This test has not been cleared or approved by the Food and Drug Administration (FDA). However, the laboratory is regulated under the Clinical Laboratory Improvement Amendments (CLIA) as qualified to perform high complexity clinical testing and this assay has been validated in accordance with CLIA standards.

This report does not represent medical advice but should be reviewed with a genetic counselor, medical geneticist or physician skilled in genetic result interpretation and the relevant medical literature. Prior consent of the patient is required to confirm understanding of the risks, benefits and limitations to this testing. Consultation with a genetic counselor is recommended pre- and post-testing.