Mosaicism Ratio: Personalized NIPT insights when you need them most

Refine each patient’s risk for aneuploidy using personalized data specific to their NIPT results.

The final result is the most important aspect of a non-invasive prenatal test (NIPT), and while most NIPT laboratories work hard to deliver the best possible results, experience, sequencing technology, and capability varies from lab to lab, along with result reliability and the risk for false positive results.

After nearly a decade of experience with NIPT, one insight that has emerged is the tendency for NIPT samples with ‘mosaic’ data to have lower positive predictive values (PPV). MaterniT 21 PLUS with Mosaicism Ratio helps to refine your patient’s risk for aneuploidy using personalized data specific to their results, building upon the current generic PPV calculation using maternal age and gestational age.

Now there is a way to identify which NIPT results are more likely to be true positive results, offering providers greater reliability and confidence as they counsel their patients.

What is Mosaicism Ratio?
- A metric calculated in the event of a positive NIPT result for trisomy 21, 18, or 13
- Requires two data points:
  - Affected fraction – the percentage of cell-free DNA that is impacted by aneuploidy
  - Fetal fraction – the percentage of cell-free DNA contributed by the pregnancy

How is Mosaicism Ratio interpreted?

Positive results are classified into one of three Mosaicism Ratio groups:

<table>
<thead>
<tr>
<th>Groups</th>
<th>Likelihood of POSITIVE result being a TRUE POSITIVE</th>
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</thead>
<tbody>
<tr>
<td>High mosaic</td>
<td>Positive NIPT result is more likely to be a true positive, associated with a higher PPV (see report below)</td>
</tr>
<tr>
<td>Low mosaic</td>
<td>Positive NIPT result is more likely to be a false positive, associated with a lower PPV (see report below)</td>
</tr>
<tr>
<td>Non-mosaic</td>
<td>Positive NIPT result that is very likely to be a true positive, associated with the highest PPV. No ‘mosaic’ language will display on the report.</td>
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In general, as Mosaicism Ratio increases, so does the positive predictive value (PPV) of the result. The PPV associated with an individual patient result depends on the trisomy involved and the Mosaicism Ratio group associated with the results (i.e. non-mosaic, high mosaic, or low mosaic).

How is Mosaicism Ratio reported? (Singleton pregnancies only)

For patients with a positive result for trisomies 21, 18, and 13, a Mosaicism Ratio comment will feature in the Lab Director Comments box on the MaterniT 21 PLUS lab report.
What are the potential causes of a low Mosaicism Ratio?

Though it may be difficult to pinpoint the exact cause of a low Mosaicism Ratio in any particular case, in general a depressed Mosaicism Ratio may be the result of placental mosaicism, prior co-twin demise, or other biological phenomenon.

Summary

- Mosaicism Ratio allows clinicians to provide customized counseling for patients with positive results for trisomy 21, 18, and 13 in singleton pregnancies
- The lower the Mosaicism Ratio, the greater the chance for a discordant result from diagnostic testing
- MaterniT 21 PLUS with Mosaicism Ratio identifies when a positive result is more likely to be a true positive
- Regardless of Mosaicism Ratio, diagnostic testing and genetic counseling are recommended for all positive NIPT results according to ACOG, Practice Bulletin 226

REFERENCES


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