How are the results reported?

- Results take 7 - 10 days to be reported to CRGH.
- Low risk results will be reported to you by letter, with a copy of your report.
- High risk results will be communicated to you by telephone. We will arrange counselling and offer further testing to see if the pregnancy is actually affected.

This screening test results are reported in 3 possible ways:

No Aneuploidy Detected:
The test identified the expected number of copies of chromosomes. This indicates your pregnancy is very unlikely to be affected by the aneuploidies tested.

Aneuploidy Detected:
The test identified too many or too few copies of one of the chromosomes. Your CRGH doctor will contact you to discuss any abnormal results and further testing options such as chorionic villus sampling (CVS) or amniocentesis, to confirm the Genesis Serenity® blood test result. Counselling will be offered for all abnormal results.

Aneuploidy Suspected:
This occurs in <0.5% of tests and simply means the test was a borderline result which was inconclusive. This could be due to many reasons for example; blood sampling or lower levels of foetal DNA. Genesis Genetics offer free of charge retest a week later in these cases if desired. It is advised to treat a suspected result in the same follow up way as a detected result.

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NIPT

What is the Genesis Serenity® Non-Invasive Prenatal Testing (NIPT)?

Prior to the Genesis Serenity® test you will require an ultrasound scan performed at CRGH to confirm your gestational dates and foetal viability. On the same day as the scan the Genesis Serenity® blood test is taken from the mother.

The blood sample is sent to the Genesis laboratory and the foetal DNA circulating in the mother’s blood is tested for:

- Patau’s syndrome (trisomy 13)
- Edward’s syndrome (trisomy 18)
- Down’s syndrome (trisomy 21)
- Sex Chromosome abnormalities such as Turner’s syndrome or Kleinfelter’s syndrome

You can also request to know the sex of your expected baby.

Who can have the Serenity NIPT test?

You might consider this test if:

- You are at least 10 weeks pregnant and would like to know if the baby is most likely to be chromosomally normal.
- Your ultrasound shows concerns or anomalies with the baby’s growth and/or development.
- Your combined test (nuchal scan & blood test at 13-14 weeks) result was high risk and you would like another test choice before committing to an invasive diagnostic procedure such as amniocentesis.
- You have personal or family history of chromosomal anomalies.

The test can be performed for all pregnant women from ten weeks of pregnancy. The test can be performed for babies conceived through fertility treatment or a natural conception.

The test can be performed accurately for ongoing single or multiple pregnancies. However, it is not recommended for twin pregnancies that have reduced to a single pregnancy.

Why test for the common aneuploidies?

Trisomies occur when three, instead of the usual two, copies of a chromosome are present. Trisomy 21, trisomy 18, and trisomy 13 are three of the most commonly occurring trisomies seen in babies at birth. Although the outcomes are variable, these conditions can cause mild to severe intellectual disabilities, and can cause multiple physical problems including congenital heart defects, defects in other organs, and a shortened life span. A foetus with a trisomy is also much more likely to be still-born or suffer from late miscarriage.

Early detection of these aneuploidies enables the early counselling of patients. The chance of having a baby with one of these conditions gets higher as a woman gets older.

Why CRGH chooses to offer the Genesis Serenity NIPT

- The overall accuracy is greater than 99.7%
- It is a simple non-invasive test consisting of a scan and a single blood sample from your arm
- It can be performed as early as 10 weeks from the start of your last period (natural conception) or from 8 weeks after the date of embryo transfer (assisted conception)
- It can test for more than only Down’s syndrome, as offered by a nuchal scan and blood test by the NHS
- Performing the test does not expose the baby to any risk
- Testing laboratories are based in London so results are returned quickly.
- Genesis provides access to licensed counselling by trained genetic counsellors

How do I know the test if effective?

Performance metrics for the test have been calculated from over 85,000 real clinical samples and includes all the false positive, false negative and predicted value rates associated with each chromosome (www.genesis-serenity.com)

<table>
<thead>
<tr>
<th>Chromosome</th>
<th>Sensitivity</th>
<th>Specificity</th>
</tr>
</thead>
<tbody>
<tr>
<td>13</td>
<td>98.15%</td>
<td>99.95%</td>
</tr>
<tr>
<td>18</td>
<td>98.31%</td>
<td>99.90%</td>
</tr>
<tr>
<td>21</td>
<td>99.14%</td>
<td>99.94%</td>
</tr>
<tr>
<td>Monosomy X</td>
<td>95.50%</td>
<td>99.00%</td>
</tr>
<tr>
<td>XX</td>
<td>97.60%</td>
<td>99.00%</td>
</tr>
<tr>
<td>XY</td>
<td>99.10%</td>
<td>98.90%</td>
</tr>
<tr>
<td>XXX/XXY/XYY</td>
<td>Other sex aneuploidies will be reported if detected</td>
<td></td>
</tr>
</tbody>
</table>

Note: Whilst the test is highly accurate, it is not 100% conclusive. A ‘No Aneuploidy Detected’ result does not eliminate the possibility of other chromosomal/genetic conditions, birth defects or other complications.

Karyotype showing Trisomy 21 (Down’s Syndrome) where rest of chromosomes have only the expected 2 copies

Performance Data Table

(Received from Illumina and Genesis Genetics data on file - available on request)