TDL Genetics are now offering 22q11.2 deletion screening as an additional option in the Harmony prenatal test menu. 22q11.2 deletion is the underlying cause of conditions described as DiGeorge syndrome and velocardiofacial syndrome (VCFS).

Why is 22q11.2 being included in the Harmony test (and not other microdeletion syndromes)?

- The 22q11.2 deletion has been carefully chosen as the only clinically relevant microdeletion syndrome to include with NIPT.
- 22q11.2 deletion is the most common chromosomal microdeletion, occurring in up to 1 in 1000 pregnancies.
- Other microdeletion syndromes have a much lower incidence and would increase the false positive rate of the test.

What is the performance of the 22q.11.2 addition?

- Inclusion of 22q11.2 deletion is aimed at a screening population, the test has been shown to identify 75% of pregnancies with a 22q11.2 deletion. Therefore, pregnancies with a known higher risk of 22q11.2 deletion, whether ascertained through ultrasound scan or family history should consider invasive diagnostic testing as this test will not identify 1 in 4 (25%) of cases.
- There is a false-positive rate of up to 0.5% associated with the 22q11.2 part of the Harmony test. This means that in 200 women with a pregnancy unaffected by 22q11.2 deletion 199 will receive a low probability result and 1 will receive a high probability result.

What is the benefit of finding out that a pregnancy has a high probability of a 22q11.2 deletion?

- Early screening and diagnosis of 22q11.2 deletions affects pregnancy management
- Following confirmatory diagnosis of 22q.11.2 deletion the following may be recommended:
  - Level II ultrasound with fetal echocardiogram to evaluate for anomalies such as congenital heart defect and cleft palate.
  - Screening for and coordinated management of associated conditions
  - Delivery at a tertiary care centre

How do I request the 22q11.2 additional test option?

- Our updated request forms include the option of selecting 22q11.2 deletion. Tick this box if this is required
- The 22q11.2 deletion cannot be requested in twin pregnancies or in pregnancies where the mother has a 22q11.2 duplication or deletion
- There is an additional charge of £75 for 22q11.2 deletion
- When discussing the informed consent for the Harmony test with your patient you must ensure they have read all the information on the reverse of the request form including the additional section headed ‘What are the limitations of the Harmony prenatal test for 22q11.2?’

If we detect a high risk pregnancy for 22q11.2 deletion we will offer a confirmatory aCGH (microarray) on the CVS or Amnio at no extra charge.

If you would like any further information about the 22q11.2 test please contact us at **TDL Genetics** by phone **020 7307 7409** or email **harmony@tdlgenetics.com**

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