Patient Information for

22q11.2 DELETION SYNDROME

The Harmony non-invasive prenatal test is based on cell-free DNA analysis and is considered a prenatal screening test, not a diagnostic test. Harmony does not screen for potential chromosomal or genetic conditions other than those expressly identified in this document. All women should discuss their results with their healthcare provider who can recommend confirmatory, diagnostic testing where appropriate.

The Harmony prenatal test was developed and its performance characteristics determined by Ariosa Diagnostics, Inc., a CLIA-certified and CAP-accredited clinical laboratory in San Jose, CA USA. This testing service has not been cleared or approved by the US Food and Drug Administration (FDA).
Why Test for 22q11.2 Deletion During Pregnancy?

Early prenatal testing for 22q11.2 deletion provides families and doctors with important information in order to properly care for the pregnancy and the newborn. Babies with 22q11.2 deletion may have special healthcare needs. Some babies with 22q11.2 deletion need surgery in the newborn period.4

Who Should Consider Prenatal Testing for 22q11.2 Deletion?

Any pregnancy has a chance to have 22q11.2 deletion. Babies born with 22q11.2 deletion usually do not have a family history of the condition.5 Unlike other genetic conditions, women of all ages have an equal chance to have a baby with 22q11.2 deletion.

What information will Harmony prenatal test with 22q11.2 provide?

Harmony prenatal test with 22q11.2 is a screening test. The purpose of a screening test is to identify pregnancies which may have an increased chance of a certain condition. If results indicate an increased chance of 22q11.2 deletion in a pregnancy, additional testing may be offered in order to confirm whether the pregnancy actually has 22q11.2 deletion.

Women who have a family history of 22q11.2 deletion or ultrasound findings which suggest the pregnancy may have 22q11.2 deletion should speak with their doctor about what testing options may be most appropriate.

Websites with more information regarding 22q11.2 deletion

- International 22q Foundation
  http://www.22q.org/
- 22q Family Foundation
  http://22qfamilyfoundation.org/
- 22q11.2 Society
  http://www.22qsociety.org/