harmony®
PRENATAL TEST

Roche

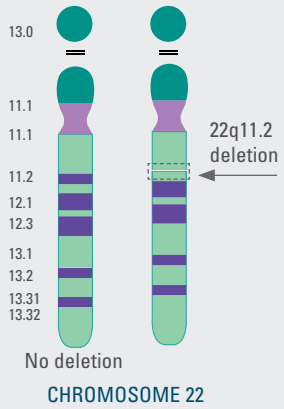
Patient Information for
22q11.2 DELETION SYNDROME

The Harmony Prenatal Test was developed, and its performance characteristics determined by Ariosa Diagnostics, a CLIA and CAP accredited clinical laboratory in San Jose, CA USA. This testing service has not been cleared or approved by the US FDA. Harmony is a non-invasive prenatal test (NIPT) 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. Before making any treatment decisions, all women should discuss their results with their healthcare provider, who can recommend confirmatory, diagnostic testing where appropriate.

What is 22q11.2 Deletion Syndrome?

22q11.2 deletion syndrome is caused by a missing piece of chromosome 22. It occurs in close to 1 in 1,000 pregnancies.[1]

The majority of children with 22q11.2 deletion have heart defects, immune system problems, learning challenges, and specific facial features. Some children with the condition also have intellectual disability, feeding problems, speech delays, low calcium levels, kidney problems, and/or seizures.[2,3] Children with 22q11.2 deletion have an increased chance of autism, and may develop psychiatric conditions later in life.[3]



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 syndrome:

International 22q Foundation
http://www.22q.org/

22q Family Foundation
http://22qfamilyfoundation.org/

22q11.2 Society
http://www.22qsociety.org/

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References
1. Grati et al. Prenat Diagn. 2015 Aug;35(8):801-9.
2. McDonald-McGinn et al. Genet Couns. 1999;10(1):11-24.
3. Bassett et al. J Pediatr. 2011 Aug;159(2):332-9.
4. McDonald-McGinn et al. Nature Reviews Disease Primer. 2015 Nov 19.
5. McDonald-McGinn et al. Genet Med. 2001 Jan-Feb;3(1):23-9.

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