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The quality choice for DiGeorge Syndrome (22q11.2 deletion) screening

22q11.2

is now part of the Harmony prenatal test menu. Early prenatal screening for 22q11.2 deletion combined with diagnosis enables informed choices and appropriate obstetrical and neonatal management.[1] Order 22q11.2 by selecting the check box on the Harmony requisition form.

Recommend the Harmony Test

Benefits of the Harmony test:

- Flexible testing options and clinically relevant testing
- Reliable timely results regardless of test options ordered
- Minimise unnecessary invasive procedures due to false-positives[2]

Choose tests that are clinically relevant to your patients, not test panels for rare microdeletions. Each condition tested has an associated false-positive rate and adds to the total false-positive rate of the test.

Performance

22q11.2 Deletion

	Detection Rate	False-positive Rate
within the 3 Mb region*	75%[3]	0.5%[3]

*including smaller nested deletions

Options for Ordering

✓ **Harmony prenatal test: Assesses the probability of fetal trisomy 21, trisomy 18, trisomy 13. Validated for use in twin and IVF pregnancies, including self and non-self donor pregnancies.[4]**

Additional Test Offerings:

- ☐ Fetal sex*
- ☐ Monosomy X**
- ☐ Sex chromosome aneuploidy panel**
- ☐ 22q11.2**

** Singletons only *Singletons and twins

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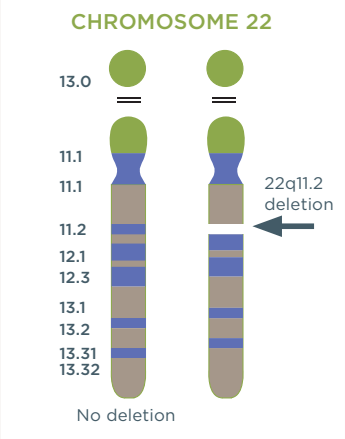
RECOMMEND THE HARMONY PRENATAL TEST

Screening Option: DiGeorge Syndrome (22q11.2deletion)



22q11.2 deletion is the most common chromosomal microdeletion[5]

This condition may occur in as many as

1 in 1000

pregnancies.[6]

It is the second most common cause of developmental delay after Down syndrome.[7]

Identify pregnancies which may be at increased risk early:

- Maternal age is not a risk factor for microdeletions[6]
- More than 90% of affected individuals have no family history of 22q11.2 deletion[8]
- 22q11.2 deletion is not reliably detected by routine screening or karyotype[7]

Clinically Relevant

22q11.2 deletion is the underlying cause of conditions described as DiGeorge syndrome and velocardiofacial syndrome (VCFS). Clinical presentation demonstrates a wide range of severity that cannot be predicted prenatally.

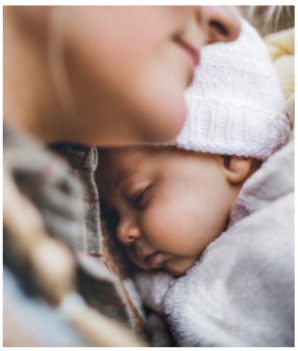
Features are diverse and may include the following:[9]

- congenital heart disease
- palatal anomalies
- immune deficiency
- hypocalcemia

Other features may include renal anomalies, learning difficulties, developmental delays, and psychiatric illness.[9]

Early screening and diagnosis of 22q11.2 deletion affects management of pregnancy. If a pregnancy is affected with 22q11.2 deletion, the following is recommended:[10]

- Level II ultrasound with fetal echocardiogram to evaluate for anomalies such as congenital heart defect, cleft palate, etc.
- Screening for and coordinated management of associated conditions
- Delivery at a tertiary care center



NIPT is a screening test. If a pregnancy is known to be at increased chance for 22q11.2 deletion based on family history or ultrasound findings, diagnostic testing should be considered.

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 prenatal test 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).

1. McDonald-McGinn et al. Nature Reviews Disease Primer. 2015 Nov 19.
2. Wax et al. J Clin Ultrasound. 2015 Jan;43(1):1-6
3. Schmid et al. Fetal Diagn Ther. 2017 Nov 8. doi: 10.1159/000484317
4. Stokowski et al. Prenat Diagn. 2015 Oct; DOI: 10.1002/pd.4686
5. McDonald-McGinn DM, Emanuel BS, Zackai EH. 22q11.2 Deletion Syndrome. 1999 Sep 23
6. Grati et al. Prenat Diagn. 2015 Aug;35(8):801-9.
7. Bassett et al. J Pediatr. 2011 Aug;159(2):332-9.
8. McDonald-McGinn et al. Genet Med. 2001 Jan-Feb;3(1):23-9.
9. McDonald-McGinn et al. Genet Couns. 1999;10(1):11-24.
10. McDonald-McGinn et al. GeneReviews (2013) http://www.ncbi.nlm.nih.gov/books/NBK1523/

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