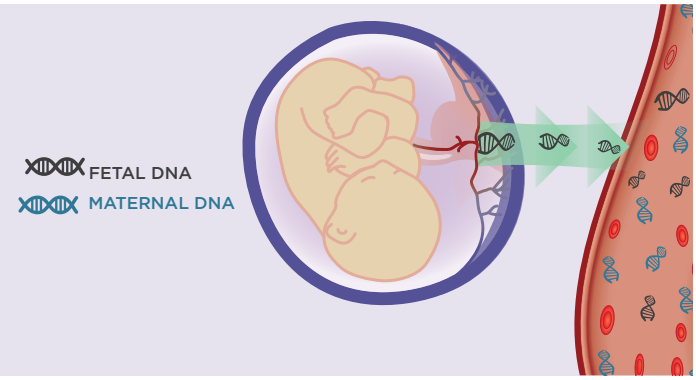
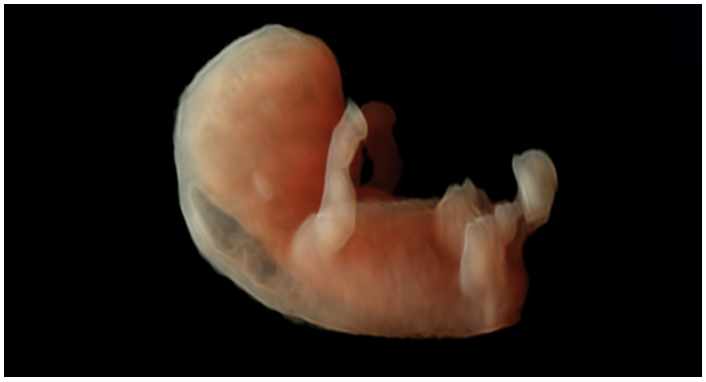
Voluson™ Ultrasound Systems Harmony® Prenatal Testing

Enabling early, focused, and efficient guidance for maternal fetal care

The 10th to 14th weeks of gestation present a unique opportunity to learn about the health of a pregnancy. Advances in ultrasound technology and maternal blood analysis have allowed for first trimester evaluation for serious fetal anomalies and more accurate screening for chromosomal conditions. A careful consideration of the roles ultrasound and cell-free DNA analysis play in prenatal screening is imperative to providing the highest level of prenatal care.

Traditionally, first trimester evaluation for chromosomal conditions was done via First Trimester Combined Screening (FTS). FTS uses a nuchal translucency measurement in combination with first trimester maternal serum analytes to evaluate for Down syndrome, trisomy 18 and trisomy 13 (the "common" trisomies). The screen performs with 80% detection rate for Down syndrome at a 5% false positive rate, which can lead to unnecessary invasive procedures.[1]





Enlarged NT at 11 weeks visualized with HDlive™ – Image courtesy of Dr. Simon Meagher

Cell-free DNA analysis screens for the common trisomies by analyzing fragments of fetal DNA in maternal blood as early as 10 weeks of pregnancy. Results do not require data from ultrasound other than gestational age, and performance varies by technology. The landmark NEXT study compared the performance of the Harmony test and FTS for Down syndrome. 38 of 38 cases of Down syndrome were detected by the Harmony test, with a false positive rate of less than 0.1%.[1]

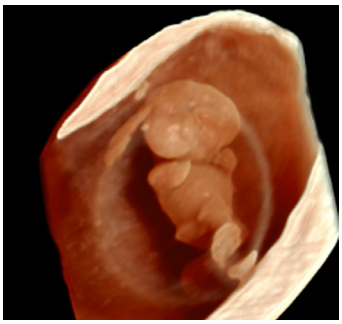
NEXT Study Results

(n=15,841)

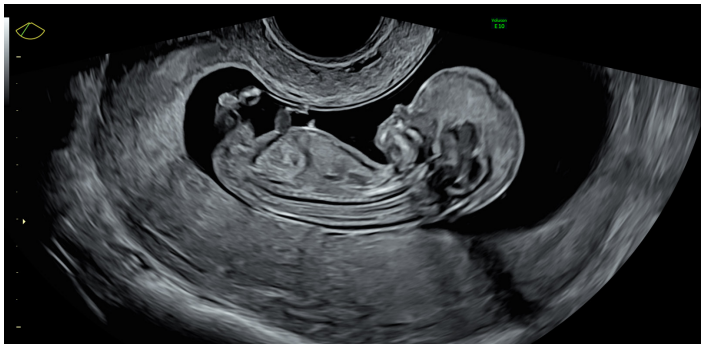
Detection Rate (affected pregnancies correctly identified as high risk)	**Harmony (38 of 38)**	100%
	FTS* (30 of 38)	79%
	p<0.001	
False Positive (unaffected pregnancies incorrectly identified as high risk)	**Harmony (9 of 15,803)**	0.06%
	FTS* (854 of 15,803)	5.4%
	p<0.001	
Positive Predictive Value (PPV) (likelihood that a positive result is confirmed on diagnostic testing, based on false-positive rate and population frequency)	**Harmony Prenatal Test**	81%
	FTS*	3.4%

The superior performance of cell-free DNA screening has led to its broad adoption as a primary screen for the common trisomies. The benefits of this approach – particularly the significant decrease in false positives and subsequent unnecessary invasive procedures – are well documented.[2-4] Professional societies recommend that all pregnant women be informed of cell-free DNA as a screening option.[5-7]

While cell-free DNA screening does not require ultrasound to produce a result, the importance of a detailed first trimester ultrasound cannot be overlooked. A detailed first trimester ultrasound can detect malformations that can either occur in isolation or with maternal or genetic disease.[8] Such findings provide early information regarding the health of the pregnancy and can lead to changes in pregnancy management. Similarly, all cases of common aneuploidies may not be detected by ultrasound alone.[9] A paradigm that uses both first trimester ultrasound and targeted NIPT can allow for early, accurate evaluation for clinically relevant conditions.



A case of Acrania at 8 weeks visualized with HDlive – Image courtesy of Dr. Simon Meagher



Stunning B-Mode image of a 13 week fetus

The complimentary technology available with the Harmony test and Voluson Ultrasound Systems can help deliver proven, clinically relevant information allowing clinicians and patients to have clear answers sooner.

harmony®

Harmony Testing

At Roche, we are committed to proven clinical science and patient care. The Harmony test evaluates the probability of trisomies (trisomy 21, 18 and 13) and additional menu options, including 22q11.2 microdeletion by analyzing cell-free DNA (cfDNA) in maternal blood. The targeted menu focuses on clinically relevant conditions and results in a low false positive rate to minimize unnecessary invasive procedures. Using the DANSR assay, a proprietary targeted DNA-based technology that focuses on cfDNA from the chromosomes of interest, and FORTE, a powerful algorithm that calculates probability by incorporating fetal fraction, the Harmony test provides accurate and reliable NIPT results.[10] To date, over 1.4 million tests have been run, and >59 peer-reviewed publications have been reported, providing clinical evidence for the Harmony test performance across any age or risk category.[11,12] The Harmony test provides high quality, clinically meaningful results so you and your patients can move on to the next step of pregnancy care.

Voluson

Voluson Ultrasound

At GE Women's Health Ultrasound, we are committed to creating a healthier future for women. Voluson ultrasound systems empower the kind of exceptional care that will exponentially improve women's lives and ensure better health for generations. By adding a detailed ultrasound around 12-14 weeks to NIPT, detection of other anomalies such as congenital heart defects, spina bifida, limb defects, congenital diaphragmatic hernia can be identified. Using Voluson ultrasound systems can help improve the detection rates of chromosomal and structural anomalies for earlier detection, providing answers sooner for better decision making.

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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 chromosomalor 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, 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.

References

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