WHAT ARE CHROMOSOMAL ABNORMALITIES?

These are abnormalities that occur in a person's genetic make-up.

We have over 30,000 genes that are stored on 23 pairs of chromosomes. These are like strands in the centre, or nucleus, of each cell in the body. **The most common chromosomal abnormality is Down Syndrome (Trisomy 21)** where there are three copies of the 21st pair of chromosomes instead of the usual two. There are other more severe chromosomal abnormalities, some of which are incompatible with life. These differ from structural abnormalities such as club feet or spina bifida.

THE RESULTS SO FAR

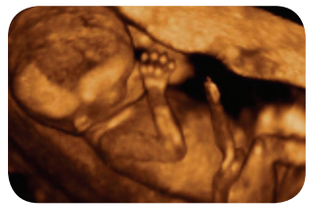
First trimester screening with ultrasound is currently offered throughout the developed world & millions of women have been examined. **Eighty percent of fetuses with chromosomal abnormalities have been identified accepting a 5% false positive rate.** This compares favourably to the detection rates achieved with screening based on maternal age alone (a 30% detection rate) and maternal age plus serum biochemistry (a 60% detection rate).

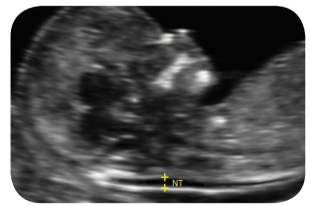
AT ASCOT RADIOLOGY, all our sonographers and reporting radiologists have undergone extensive training in all aspects of nuchal scanning. We have been carrying out these examinations since we first opened in April 1999.

We were one of the first private radiology practices in New Zealand to be accredited in nuchal scanning and have all the most up to date fetal medicine software to allow analysis.

FURTHER INFORMATION on obstetric ultrasound, including **fetal anatomy and 4D scanning,** and a current price list, can also be found on our website:

www.ascotrad.co.nz/Our-Services/Ultrasound





NUCHAL SCANS, and all other obstetric ultrasound scans, are available at these Ascot Radiology branches:

ASCOT HOSPITAL: 90 Green Lane East, Remuera
TEL: (09) 520 9550 or 0800 ULTRASOUND (0800 858 727)
EMAIL: us@ascotrad.co.nz

AUT MILLENNIUM: 17 Antares Place, Mairangi Bay
TEL: (09) 478 6640 EMAIL: millennium@ascotrad.co.nz

BIRTHCARE, PARNELL: 20 Titoki Street, Parnell
TEL: (09) 373 5988 EMAIL: parnell@ascotrad.co.nz

ST HELIERS: Level 1, 8-10 Turua Steet, St Heliers
TEL: (09) 555 9558 EMAIL: stheliers@ascotrad.co.nz

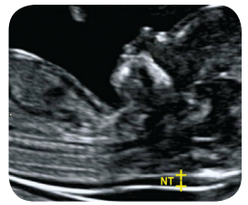
TO MAKE AN APPOINTMENT,
Please call us during office hours,
8.30am - 5pm Monday to Friday,
or go to our website:
www.ascotrad.co.nz/Book-A-Scan

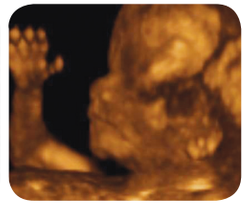
ASCOT RADIOLOGY
www.ascotrad.co.nz

SOME OF THE ULTRASOUND IMAGES USED IN THIS BROCHURE ARE COURTESY OF GE & PHILIPS HEALTHCARE

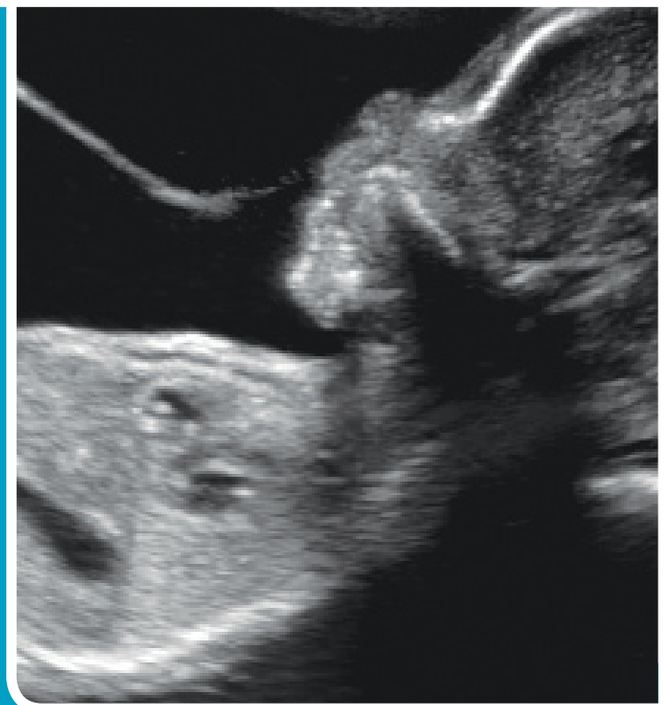
ASCOT RADIOLOGY

OBSTETRIC ULTRASOUND





THE 12 - 13 1/2 WEEK CHROMOSOMAL ASSESSMENT SCAN



ASCOT HOSPITAL • AUT MILLENNIUM
BIRTHCARE, PARNELL • ST HELIERS

ABOUT NUCHAL SCANNING

- Every woman has a chance of having a baby with Down Syndrome. As you get older, the chance is increased.

- Your chance is the same as anyone else of the same age. **(See chart below).**

- **The nuchal scan is a screening test for Down Syndrome and other chromosomal abnormalities.** You will not get an absolute answer about your baby's chromosomes from this scan.

- **Our measurements are combined with results from your blood test to give you an adjusted chance of having a baby with Down Syndrome.** This is called a Maternal Serum Screen 1 (MSS1). This takes into account your age, your blood test results, and the nuchal fold measurement. The nuchal fold is a measurement of the skin thickness behind the baby's neck.

This adjusted chance often helps couples decide about proceeding with further tests such as an **amniocentesis or Chorionic Villus Sampling (CVS).** These are both more invasive tests but they do give absolute answers.

AGE (YEARS)	CHANCE OF TRISOMY 21	
	AT BIRTH	AT 12 WEEKS
20	1 in 1526	1 in 1018
25	1 in 1351	1 in 901
30	1 in 894	1 in 596
32	1 in 658	1 in 439
34	1 in 445	1 in 297
36	1 in 280	1 in 187
38	1 in 167	1 in 112
40	1 in 96	1 in 64
42	1 in 55	1 in 36

For instance, if your chance after the nuchal scan and blood test was very low, you may decide against having an amniocentesis or CVS. However, if your chance was high (1 in 300 or greater) you may wish to carry on with further testing.

Please note that most amniocentesis and CVS results are normal.

HOW DOES NUCHAL SCANNING WORK?

- **The best time to come for a nuchal scan is from 12 weeks until 13 1/2 weeks** as this increases the accuracy.

- The sonographer will take measurements during the scan to ensure the baby is growing appropriately for dates.

- **A few measurements of the nuchal fold will be made to get as accurate measurement as possible.** We are measuring a very small structure (usually less than 3mm) so the baby has to be in an optimum position. Often you will be asked to roll, cough or sit up to help the baby move into a better position. Occasionally you may have to fill your bladder to help us visualise the baby.

- **A brief scan of the baby's anatomy will also be carried out.** A more comprehensive scan will be done at 18-20 weeks gestation.

- **The measurements will be forwarded to the lab to be combined with the results of your blood test.** A copy will also be sent to your lead maternity carer (LMC). You will get your overall results at your next appointment with your LMC.

- **However, if your chance is high, or if we are concerned about your scan,** we will make you aware of this before your leave the department so you can obtain your results from the lab more promptly.

Only about 80% of chromosomally abnormal babies have thickened nuchal folds. Some look normal which is why, as with all screening tests, some normal babies are identified as abnormal (false positives) and some abnormal babies are identified as normal (false negatives).

If you have any questions during the scan, please feel free to ask the sonographer or radiologist.

NON INVASIVE PRENATAL TESTING (NIPT)

There is a new screening option for Down Syndrome called Non Invasive Prenatal Testing (NIPT). This provides a higher level of accuracy when compared to nuchal scanning and MSS1 (the routine blood test). It is a user pays option and at Ascot Radiology, we offer this in conjunction with counselling, as recommended by the New Zealand Maternal Fetal Medicine Network (NZMFMN).

For more information on NIPT, please go to our website: www.ascotrad.co.nz/Our-Services

AMNIOCENTESIS & CHORIONIC VILLUS SAMPLING

To get an absolute answer with regard to your baby's chromosomes, you will need to have another test – **either a Chorionic Villus Sampling (CVS) or an amniocentesis.** These tests carry a small chance (1 in 500 to 1 in 1000) of causing miscarriage and are not usually recommended unless you have a high chance from a screening test (1 to 300 or greater on MSS1). Your LMC can discuss these tests with you and help you reach your decision.

CVS: This is done from 11 1/2 weeks up to 14 weeks gestation. The skin of the abdomen is numbed with local anaesthetic and a very fine needle is passed into the uterus through the mother's abdomen using ultrasound as a guide. A sample of the chorionic villi is taken from the placenta.

Amniocentesis: This is done at 15 to 17 weeks. A very fine needle is passed into the uterus through the mother's abdomen using ultrasound as a guide. A sample of the amniotic fluid is taken from around the baby.

Both tests obtain cells that are then grown to check for any abnormality. The results can be expected in approximately 2 weeks.