Obstetric and Gynaecological Ultrasound in Mulgrave (formerly Diagnostic Women's Ultrasound), has provided specialist pregnancy and gynaecological ultrasound services to Melbourne's South East and the Dandenong region since 1989.

Dr Simon Meagher is an obstetrician/gynaecologist who has sub-specialised in ultrasound and prenatal diagnosis. He is the Director of Monash Ultrasound for Women, consultant staff specialist at the Mercy Hospital for Women and lecturer at both Melbourne and Monash Universities. He is well known for his clinical and procedural skills and academic achievements, having over 40 publications in local and international journals.

O & G Ultrasound provides a complete range of pregnancy and gynaecological ultrasounds and related procedures including prenatal genetic testing for Down Syndrome. Dr Meagher (obstetrician/ gynaecologist/ultrasonologist), Associates and sonographers specialised in obstetric ultrasound, work with state of the art, high resolution ultrasound machines. Together with an experienced team of nurses and support staff, our aim is to provide you with the highest quality ultrasound service available.

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OBSTETRIC & GYNAECOLOGICAL ULTRASOUND

Down Syndrome

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What is Down Syndrome?

Down Syndrome is one of the most common genetic disorders which gives rise to both physical and mental abnormalities in the baby and growing child. The typical abnormalities vary but include:

1. Short stature
2. Abnormal facial features
3. Heart abnormalities such as a hole in the heart
4. Bowel abnormalities such as bowel obstruction
5. Kidney abnormalities.
6. Mental retardation

The facial features of Down syndrome babies resemble the Mongolian race and hence the term "Mongoloid". Down syndrome children have an intelligent quotient (IQ) which is well below the normal population.

What causes Down Syndrome?

Down Syndrome is where there is an numerical abnormality of the chromosomes. Chromosomes are the building blocks of the human body and are present in every human cell. Normally there are 23 pairs of chromosomes, (46 in total) with each chromosome determining a specific feature such as height, eye colour, hair colour, finger size, etc. The developing baby inherits 23 chromosomes from its father and 23 from its mother. The Down Syndrome chromosome is No. 21. In normal situations the baby will receive one copy from each parent. If however during the meeting or fusion of the sperm and egg the baby receives an extra copy chromosome 21 then every cell will contain three copies and the total count in each sell will be 47. This is called Trisomy 21 or Down Syndrome. It is not known why the extra chromosome is inherited but we know that the chances of this happening increases with the advancing age of the mother.

What is the long term outlook for babies with Down Syndrome?

This varies considerably and depends on the number and degree of physical disabilities. Many of the structural abnormalities such as heart and gut defects are correctible with surgery. Overall however the life span of Down Syndrome children is reduced.

Where can I find out more information about Down Syndrome?

More information may be obtained from the Down Syndrome Association of Victoria (Tel : 03.94862377) or from the internet http:www.downsyndrome.com.

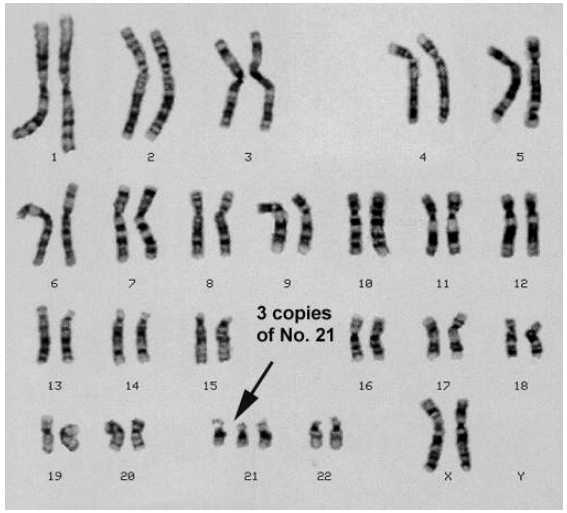
What Down Syndrome tests are available to me and my unborn baby?

There are **screening tests** (triple test, quadruple test, & ultrasound) which estimate your level of risk and may identify up to 80% of Down Syndrome fetuses and there are **diagnostic tests** (amniocentesis & CVS) which identify 100% of Down Syndrome pregnancies. Screening tests carry no risk to the pregnancy but diagnostic tests increase the background risk of miscarriage by 0.5-1%.

The screening tests include an ultrasound at 11-14 weeks (70-80% detection rate), blood tests at 15-20 weeks (70% detection rate), ultrasound at 18 weeks (40% detection rate). More recently the 12 week ultrasound and a 9-10 week blood test have been combined to give up to a 90% detection rate of Down Syndrome. It must however be appreciated that these tests are not definitive and whilst they may detect between 70-90% of Down Syndrome it implies they will miss 10-30%.

The definitive tests available include needle tests such as amniocentesis, CVS or fetal blood sampling. Both amniocentesis and CVS involve direct sampling of the babies cells and examination of the chromosomes (refer patient information leaflet on CVS and amniocentesis). Both tests carry a small risk of miscarriage which needs to be considered. You may discuss these tests in more detail with your family doctor or specialist.

Detailed analysis of the chromosomes



A cell from the baby obtained at amniocentesis is split open and examined. In this baby there were 3 copies of the number 21 chromosome. The baby therefore has Down syndrome.