The 12 week ultrasound:

By undersigning this document I understand the following about the 11 – 14 week ultrasound:

THE 12 WEEK ULTRASOUND HAS SEVERAL PURPOSES

1. Confirming / determining gestational age (age of the pregnancy)
2. Confirming single or multiple pregnancy
3. Confirming viability of the pregnancy (is the fetus living)
4. Confirming position of the pregnancy to be intrauterine (in the womb).
5. Determining risk of genetic abnormalities – specifically Down syndrome.

DOWN SYNDROME

Down syndrome (DS) is a genetic condition that happens at time of conception. All normal human tissue cells have 23 pairs of chromosomes, with a pair of sex chromosomes (for females this is 2 X chromosomes, and for a male an X and a Y chromosome). In the egg cell and sperm cell there is just one set of chromosomes – thus 23X in the egg cell, and 23X or 23Y in the sperm cell.
Trisomy is a condition where (usually) the egg cell miss-divided, and ended up with both chromosomes of a specific pair. In the case of Down, it is 2 copies of the chromosomes number 21. When this egg cell then fuse with the sperm cell it has 3 copies of the chromosome 21, and this leads to the typical picture noted in people with Down's. In medical terms this is called Trisomy 21 (= 3 of chromosome 21). Likewise we see Trisomy 18 & 13, typically with even more devastating results in terms of brain and other organ development. Important to take note that these conditions are far less common than Down syndrome.

Children and adults with Down syndrome share the following characteristics. A person might display all or only some of these, and in varying degrees:

Head and neck

- Up slanting eyes
- Flat facial profile/flat nasal bridge
- Folded, low set small ears
- Small head
- Open mouth with a protruding tongue
- Short neck with excessive skin at nape of the neck

Extremities

- Short broad hands
- Transverse palmar crease
- Space between the first and second toes (sandal gap)
- Hyper flexibility of joints

Patient Name:_____ Patient Signature:_____ Date:_____

Witness Name: _____ Witness Signature: _____ Date: _____

Intellectual disability (mental retardation)

Almost all individuals with DS have cognitive impairment, although the range is wide. Most are mildly to moderately intellectually disabled, with IQ in the 50 to 70 or 35 to 50 range, respectively, although some are severely impaired with IQ 20 to 35

Developmental impairment becomes apparent in the first year of life. In general, the average age of sitting (11 months), crawling (17 months), and walking (26 months) is twice the typical age. The sequence of language development is the same, although the rate is slower, with the average age for the first word at 18 months. The child with DS continues to learn new skills. However, IQ declines through the first 10 years of age, reaching a plateau in adolescence that continues into adulthood.

Behavioral and psychiatric disorders

Behavioral and psychiatric disorders are more common in DS than typical children, but less common than in those with other causes of mental retardation. Disruptive behavioral disorders, such as attention deficit hyperactivity disorder, conduct/oppositional disorder, or aggressive behavior, are most common.

Heart disease

Approximately one-half of individuals with DS have congenital heart disease.

Short stature

Growth rate is reduced in DS compared with typical children, especially in infancy and adolescence.

Obesity

The prevalence of obesity (defined as a body mass index >27.8 kg/m2 in adult males and >27.3 kg/m2 in adult females) is greater in DS than in the general population.

Eye problems

Ophthalmologic disorders that require monitoring and intervention affect the majority of children with DS.

Hearing loss

Hearing impairment affects 38 to 78 percent of individuals with DS

Endocrine (hormonal) disorders

Endocrine abnormalities in DS include thyroid dysfunction and diabetes.

Over and above these there are many other health issues affecting people with Down syndrome.

Patient Name:_____ Patient Signature:_____ Date:_____

Witness Name: _____ Witness Signature: _____ Date: _____

WHY SHOULD I SCREEN FOR DOWN SYNDROME?

- In the United States in 2002, the prevalence of Down syndrome was calculated to be 1 in 629 births when no testing and/or offering termination of pregnancy was available. The prevalence of Down syndrome is relatively high, but depends on the maternal age range of the population. The prevalence in the population increases as the proportion of pregnancies in older women increases. The prevalence of Down syndrome is highest early in pregnancy and declines naturally (through spontaneous miscarriage) throughout the pregnancy
- Down syndrome carries a significant burden of disease: the syndrome is associated with morbidity and mortality in affected individuals and high financial and psychosocial cost to their families.
- Diagnostic tests that detect the chromosomal abnormality are readily available.
- For couples who choose to prevent birth of an affected infant, safe and effective options are available.
- For families who want the opportunity to plan for the birth of an affected child, various Down syndrome associations offer support for families and individuals with Down syndrome and promote their participation in society.

WHAT DOES "SCREENING" AND "DIAGNOSIS" MEAN?:

Screening for Down syndrome means we look at the age of the mother, and according to established tables know what risk she is for having a baby with Down syndrome. Typically at 12 weeks the risk for a 20 year old is close to 1:1 000, at 30 it is 1:530, at 35 1:210, and at age 40 1:60. This means if we for example have 210 women, all aged 35, and all of them 12 weeks pregnant, one of them would have a baby affected by Down syndrome. With the screening tests we try to look at other parameters than just age, adding all of these into an algorithm and determining a new risk that could be much lower i.e. 1:3 000 or much higher i.e. 1:50. So this is not a 'yes' or 'no' answer.

This risk assessment will assist the parents and myself in making a decision regarding further testing. Further tests are called **invasive testing**. This means obtaining fetal cells through an amniocentesis (drawing fluid out from around the fetus) or alternatively a placental biopsy called CVS. Unfortunately these procedures carry some risk of miscarriage (1:200 to 1:100) as well as possible premature delivery later on. For this reason the medical consensus is to view the results as "high risk" when the risk is more than 1:300.

This also unfortunately means that some babies with Trisomy will not be diagnosed, as the risk of miscarriage outweighs the risk of Down's.

Patient Name:_____ Patient Signature:_____ Date:_____

Witness Name: _____ Witness Signature: _____ Date: _____

These procedures will give a definite answer (**diagnosis)** in 99% of cases – either being normal, or confirming Trisomy 21 (or other syndromes). Initial results with a 95% accuracy are usually available after 72 hours, and final results can take up to 6 weeks to come back.

It is necessary to understand that even when the risk is 1:50 it means 49:50 of these babies will be normal. The same goes when we say 1:4 000 – one of those 4 000 will be affected (and thus 20 babies will be miscarried in order to diagnose this one with these odds).

SCREENING FOR TRISOMY 21 AT 12 WEEKS:

During the 12 week scan some or all of the following will be checked:

- Size of baby CRL or Crown Rump Length – need to be 48 – 79mm
- Measurement of the neck skin fold (Down babies are more swollen than normal babies due to the increase chance of heart defects, as well as a connective tissue condition keeping fluid in the skin). Measurements above 3mm are a cause for concern, although 85% of these babies will still be chromosomally normal.
- Noting the presence of the nasal bone – Down babies usually have very flat nose-bridges, and only 3% of Down's babies will have a visible nasal bone. This is a great structure to identify, but it is important to note that up to 15% of normal babies will not yet have a nasal bone visible at 12w. The nasal bone, as well as angle of the nasal bone in relation to the forehead is noted.
- Ductus venosus flow – blood flow from the placenta through the umbilical vein, into the belly button, and through to the liver. The ductus venosus shunts some of this oxygen rich blood directly to baby's heart, and assists with oxygen delivery to the body. This structure closes off minutes after birth. Blood flow in the ductus venosus gives a very specific pattern in babies with normal heart development, with reversed flow due to various possible abnormalities in the heart in babies affected by Down syndrome as well as certain heart defects.
- An overview of baby can be obtained – looking for severe and obvious defects such as an absent top part of the skull, limb defects, cystic hygroma (massive swelling over the neck, associated with a missing chromosome X),and some others.

BLOOD TESTS WITH THE 12 WEEK ULTRASOUND:

Blood tests looking at certain proteins and hormones linked to the pregnancy will be incorporated at the time of the ultrasound. The tests can be done on the day of the measurements, or from 10 weeks onwards. The test has a slightly better predictive value if it is done earlier.

I prefer to have the blood tests done at around 10 weeks, and then do the measurements around 12 weeks. The final results will take about 48 hours to come back, and will again be expressed as a risk i.e. 1: xxx (with risk <1:300 being "low risk", and >1:300 being "high risk").

Patient Name:_____ Patient Signature:_____ Date:_____

Witness Name: _____ Witness Signature: _____ Date: _____

Although we suggest invasive testing with a risk higher than 1:300 the parents should be very much a part of this decision-making. Things to consider would be whether they want to do the testing at all, accepting that no diagnosis is possible, or doing the screening, but only opting for invasive testing if the risk is more than 1:100. **The toughest decision of all would be whether or not to continue with the pregnancy if Trisomy 21 has been confirmed on amniocentesis.**
These decisions are always very difficult: I and my practice staff will be available at all times to discuss these without ever leading any decisions, or judging any chosen route parents will take.

THE "NON-VISIBLE" BENEFITS OF THE 12 WEEK SCAN:

This document is not all about doom and gloom.
At 12 weeks your baby will be clearly visible as a true tiny human being. At this stage all structures have developed, baby will be able to jump and make turns and rolls while we are watching the screen.
Listening to the fetal heart for the first time is awe inspiring – and to imagine this heart will be pumping (all going according to plan) for the next 80 – 90 odd years!
We will detect certain structures – the stomach bubble confirming baby is able to swallow fluid and thus confirming that the brain, nerves and muscles are all connected to be able to do this.
The bladder is often visible, confirming that the kidneys are able to deal with fluids (the mother will be dealing with all waste products until after baby is born).
If baby cooperates we might be able to guess the sex – although this is not a guarantee!!

Most of all this and every other ultrasound could assist in the bonding of both parents and even siblings to this new little person– and this cannot be weighed or measured.

NOTES:_____
_____
_____
_____
_____
_____
_____

Patient Name:_____ Patient Signature:_____ Date:_____

Witness Name: _____ Witness Signature: _____ Date: _____