

Patient information on soft markers

Before you read this section remember the following important points.

The vast majority of babies with soft markers are normal.

Soft markers are frequently seen in healthy babies

What is a soft marker?

A soft marker is something which may be seen at the time of the 18-22 week scan which may indicate an increased chance that your baby has Down's syndrome or another chromosome anomaly but which in itself is probably of little or no significance.

What does having a soft marker mean?

Having a fetus (baby) with a soft marker means that there is a slight increase in the chance of that baby having a chromosome anomaly, for example Down's syndrome.

How high is that risk?

The risk is dependent on a number of factors these includes which soft marker has been seen, if it is seen on its own and what is your prior risk of having a baby with downs syndrome based on your age and history.

How are soft markers discovered?

Soft markers have been discovered in three ways, the first is by chance or a "eureka" moment, the second is by going back to a number of ultrasound images of babies who where born with the problem and seeing if anything unusual can be spotted. Finally we can look at a child born with a condition and then look for the characteristic features seen in children with that disorder to see if we could pick them up on ultrasound.

How can something which appears to be a problem one minute suddenly become irrelevant?

Soft markers are common and will frequently be seen on a 20 week scan. In fact about 1 baby in 30 will have a soft marker. The vast majority will not cause any problem at all. However, what has been observed in the past is that that particular marker has been seen more frequently in babies with say Down syndrome than in the "normal" population. This has allowed us to calculate the risk of individual's baby having Down syndrome. If after this calculation they opt to have an invasive test and this is reported as normal then the finding becomes irrelevant. – Because soft markers are frequently seen in normal babies.

Does that mean I have to have an invasive test?

No the decision to have an invasive test has to be based on a number of factors including your wishes, your background risk and how much that risk is increased by the particular soft marker. Invasive tests carry a risk of miscarriage and you therefore have to balance that risk against the risk of you baby having a problem. You also need to think about what you would do with the information. If for example you would never consider terminating the pregnancy then you might not want to run the risk of an invasive test

What is the relevance of specific soft markers?

Below is a list of the more common soft markers identified and a discussion of their relevance

- **Nuchal pad**

At the time of the anomaly scan we can measure the thickness at the back of the baby's head (This is different from the nuchal translucency measurement that maybe performed at 11-14 weeks). Like any measurement this can vary widely depending on many factors. It has been noticed that an increased thickening has been seen slightly more frequently in babies with chromosome anomalies, such as Down syndrome. If your baby has been noticed to have this thickening then you will be offered the opportunity to undergo a diagnostic test (either an amniocentesis or a chorion villus biopsy) to find out for sure if your baby has this problem or not. The chance of your baby being affected depends on a number of factors which include:

- Your starting risk – The younger you are the lower the risk
- How thick the nuchal pad is - An 8mm nuchal will carry a greater risk than one of 6.1mm
- Whether there are other signs suggestive of a problem or if this is an isolated feature.

- **Choroid Plexus Cyst (CPC)**

Inside our heads we have 4 fluid filled cavities called ventricles. These chambers are joined together by little tubes and allow fluid to circulate around our brain and down our spinal cord. The fluid is produced by two glands called the choroid plexus which sit in two of these ventricles one in each half of our brain. It is important to remember that the gland sits inside a fluid filled reservoir and not in a thinking part of the brain. Any gland which produces fluid will from time to time trap fluid within itself. When this happens it will produce a small cyst. In this case cyst is no more than a small collection of trapped fluid which ultrasound is very good at detecting. Early in pregnancy they are very common. At the time of the 20 week scan they will be seen in nearly one in every 50 babies. Due the way the choroid plexus develops most will have disappeared by 28 weeks. The cyst is of no functional significance. They will not do your baby any harm. Indeed it is thought that we are constantly producing these cysts in our choroid plexus throughout our lives. The only reason we ever report these cysts is because they are seen slightly more frequently in a chromosome anomaly called Trisomy 18 or Edward's syndrome. To date evidence in the literature would suggest that the risk of a fetus having Edwards syndrome based on the

finding of an isolated CPC is 1:189. This risk rises exponentially if other structural anomalies are found.

A better way of looking at the risk if the cyst is found in isolation is to increase the prior risk, based on the maternal age by a factor of 8.6.

Sadly Edwards syndrome is very much worse than Down's syndrome and most baby's die during the pregnancy or early.

There is no association between CPC and Down's syndrome.

- **Echogenic bowel.**

The finding of echogenic bowel means that the person scanning you feels that your baby's bowel is whiter than it would normally be. This is very subjective and it has been shown that different people call different degrees of brightness "echogenic" and the same person will not necessarily call the same degree of whiteness "echogenic" on two different occasions which can obviously cause problems. The most common cause appears to be the presence of blood within the amniotic fluid. This probably occurs as a result of a silent bleed during the pregnancy although you may have noticed some vaginal bleeding at some time. The baby appears to swallow this blood which then sits in the bowel making it appear bright. For the vast majority of babies this causes them no harm and is therefore of no significance.

Despite the difficulty outlined above it has been found that echogenic bowel can be found in association with a number of problems in the baby.

- Down's Syndrome
- Cystic Fibrosis
- Viral Infections
- Bowel Obstruction
- IUGR (small babies that struggle to grow in the womb)

The risk of Down's syndrome (see separate patient information sheet) is ~1-2% and the risk of Cystic Fibrosis (see separate patient information sheet) is ~2%. Babies can acquire a viral infection whilst still in the womb. In order for this to occur, as a general rule, the mother has to acquire the infection during the pregnancy. For many the placenta acts as a barrier and prevents the spread of the infection to the developing baby but for others this is not the case. Most viral illnesses like chest infections and flu do not cause a problem both others such as CMV (see separate patient information sheet) can cause harm to the baby. The chance of a viral illness being the cause of the bright bowel is in the order of 1%. We can check for this by taking blood from the mother. This result usually takes about 10 days. There is an association between bright bowel and babies whom fail to grow as well as we might expect. In our experience if this is going to be the case the baby is usually already small at the time the diagnosis of bright bowel is made. However it is likely that you will be offered another scan to check that your baby is growing at the correct rate. Finally we are aware that very occasionally this can be a marker of a bowel obstruction. Our bowel is a long pipe and if it becomes kinked or if there has been a failure in development for the hole in the middle, the

lumen, to have been formed then the bowel will be obstructed. The net result is that the bowel before the obstruction increases in size as it tries to overcome the blockage and the bowel the other side becomes smaller as it is not being used. Before birth we do not use our bowel much so this is not a major concern but may mean an operation after birth. This is a complication seen in less than 1% of cases.

In conclusion the vast majority of babies with bright bowel turn out to be normal with no long term problems. However there is a small risk of a chromosome problem (1/2%), CF 2%, a viral cause (>1%), a blockage in the bowel (>1%) and severe growth restriction (>1%). We will discuss all of these with you and offer the opportunity of having an invasive test, blood tests from the mother and father and a further ultrasound scan. It is up to you what if any further tests you opt to have. But remember although the list of potential problems is long 95% or 19 out of 20 babies are born without any problem at all.

- **Renal dilatation**

Mild renal dilation is usually defined as a renal pelvic dilatation of between 5/6 – 10 mm at the time of the mid pregnancy scan carries a small increased risk of Down's syndrome. However this risk is not felt to be of sufficient impact to alter a woman's age related risk (LR 1.9). As such we would not mention it to you unless it may occasionally mean something else. For the majority of babies this is a normal finding, however it can be a marker of two long term kidney problems reflux and a PUJ obstruction (**p**elvi**u**reteric **j**unction obstruction). The significance of these is that in the former, an infection in postnatal life, within the bladder will be refluxed back to the kidney. A PUJ obstruction if severe will lead to back pressure on the fetal kidney and can cause significant damage. This is unlikely to occur when the renal pelvic dilatation is under 15mm and highly unlikely with a diameter of 10mm or less. The chances of either of these conditions affecting your baby are small however we will usually arrange a further scan at about 32 weeks to check that it has not got any worse. If you have been told your baby's kidneys are dilated then it is worthwhile mentioning this to the baby doctor who examines your baby after delivery so that can plan a follow up appointment at the correct time.

- **Echogenic foci in the fetal heart**

Echogenic foci in the fetal heart are thought to arise as a result of calcification within the papillary muscle. They appear to be of no short or long term clinical significance other than a very weak association with Down's syndrome. We see these so often that we do not believe them to be of great significance and in the absence of any other abnormal features in your baby we would not offer any further testing.

Advice from the national screening committee re soft markers

The NSC has agreed that the following soft markers found in isolation in a women who has already undergone biochemical screening should be discounted:

- Head shape
- Choroids plexus cyst
- Sandal gap
- Two-vessel cord
- Cisterna magna
- Clenched hand
- Clinodactyly
- Echogenic foci in heart
- Short humerus

This of course does not answer the question of what should be done if the woman has not undergone biochemical screening or if more than one marker is seen. In that situation each case should be taken on its merits. It is very hard to be didactic as the risks would depend on which markers had been seen and the a priori risk based on the maternal age. For example a thickened nuchal pad and echogenic bowel in a 20 year old might be of greater significance than a slightly short femur and 5 mm renal pelvic dilatation in a 35 year old.