



RISK ASSESSMENT

Most children with serious abnormalities are born into families without a history of any disorder. Hence the need for risk assessment.

Multiple maternal blood markers and ultrasound estimate Down's syndrome and Edwards' syndrome risks. Those with high risk may consider CVS or amniocentesis. Down's syndrome risk over 1 in 250 is regarded as high.

Gene faults found in DNA, before or during pregnancy, show that someone is a carrier. When both partners are carriers there is a very high, 1 in 4, risk of an affected child. Female fragile X syndrome carriers are at high risk regardless of their partner.

MULTI-MARKER SCREENING

NHS Some multi-marker screening is now available. Our **Genmark** and **Addmark™** use additional markers to increase risk accuracy.

Down's syndrome The most common cause of severe learning disability. There are associated medical problems: but many are healthy and have a good life span.

Edwards' syndrome Ten-times less common than Down's syndrome; nearly all affected die in early infancy.

Spina bifida Failure of the neural tube to close causes physical disability but generally no mental handicap.

Ultrasound alone The main marker is nuchal translucency at 11-13 weeks or nuchal skinfold at 14-20 weeks. Many centres also determine nasal bone.

Genmark Combines 4 blood markers at 9-20 weeks with ultrasound at 11-20 weeks.

Addmark™ Initially 4 blood markers at 9-13 weeks and ultrasound at 11-13 weeks. If risk is borderline, 4 more blood markers tested after 14 weeks and risk revised.

Risk accuracy Our additional markers lead to the greatest percentage of Down's syndrome pregnancies with high risk ('detected') and the smallest percentage of pregnancies with high risk ('positive').

Test [†]	Week	Detected [‡]	Positive
Genmark	9-13	91%	1.2%
	14-20	80%	2.7%
Addmark™	9-13	93%	0.6%
Ultrasound only	11-13	87%	1.8%
	14-20	61%	3.0%

[†]When nasal bone is one of the ultrasound markers

[‡]Also, Edwards' syndrome about 90% if 9-13 weeks; spina bifida about 75% if 15-20 weeks

Maternal age The above table gives overall rates for UK women. Rates increase with age and these can be found on our website.

Nasal bone omitted Some centres do not determine nasal bone and occasionally the fetal position obscures it. Detection is lower: **Genmark** 84% and 76%; **Addmark™** 89%; ultrasound only 76% and 52%.

Ultrasound unavailable At 14-20 weeks a 4 marker blood test can be done; phone us.

Twins Detection is lower; see our website.

Multi-marker screening

- ◆ Down's syndrome
- ◆ Edwards' syndrome
- ◆ Spina bifida

DNA screening

- ◆ Fragile X syndrome
- ◆ Spinal muscular atrophy
- ◆ Cystic fibrosis
- ◆ Eight Jewish disorders

DNA SCREENING

Fragile X syndrome (FXS) Second common cause of severe learning disability. Boys more intellectually impaired than girls.

Spinal muscular atrophy (SMA) Debilitating disorder that destroys nerve cells. Main genetic cause of death in infancy.

Cystic fibrosis (CF) Excessive amounts of thick mucus in the lungs and digestive tract. Average life expectancy 30-40 years.

Jewish disorders In addition to Tay-Sachs disease there are 6 common disorders as well as specific DNA faults in CF.

Tests Carrier status for one or more of FXS, SMA and CF can be determined in a single blood sample. **Alef8** is our test for all 8 Jewish disorders.

Accuracy The rate of carriers in the population ('frequency') and percentage of carriers with identifiable DNA faults ('detected') are high.

Test	Frequency	Detected
FXS	1 in 150	>99%
SMA	1 in 60	90%
CF	1 in 25	>80%
Alef8	1 in 6 [‡]	94-99%
Tay-Sachs only	1 in 25	94%

[‡]At least one of the eight Jewish disorders

Couples Testing for SMA, CF or Alef8 can be done for couples or individuals. For couples the woman's sample is tested first and the man's sample is tested only if the woman is found to be a carrier.

PROCEDURES

If you do not already have one, the first step is to obtain a blood sampling pack. For those requiring multi-marker screening the second step is to arrange for an ultrasound scan.

Sampling pack Contains blood vials, clinical detail forms, instructions for payment and postal packaging.

Packs can be obtained from us by phone or email, and some GPs and hospitals keep stocks. If only a multi-marker screening pack is stocked and DNA screening is required, additional vials can be obtained from us.

Ultrasound appointment There is a network of ultrasound centres affiliated to **genome** including Leeds Screening Centre. Contact us for the phone number of your nearest centre or to make an appointment at Leeds Screening Centre.

Gestation We recommend that multi-marker screening is performed as early in pregnancy as possible, to allow plenty of time for further decision making.

For both **Genmark** and **Addmark™** it is best to take the blood sample 1-2 weeks before the ultrasound appointment so that a risk can be given at the time of scan.

genome offers a range of screening tests, including those previously carried out at the University of Leeds since 1991, under the direction of Professor Howard Cuckle, a leading international authority on screening.

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