

eurocat

european surveillance of
congenital anomalies

Special Report: Special Report:

Prenatal Screening Policies in Europe

2010

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Edited by Patricia Boyd and Ester Garne

Introduction

Since the setting up of EUROCAT in 1980 there has been a continuous increase in the proportion of congenital malformations that are diagnosed prenatally. EUROCAT studies have shown significant regional differences in prenatal detection rates in Europe.

A range of policies have been developed in different countries and in different areas within countries. The availability of different resources, termination of pregnancy laws and social and cultural factors are important issues which vary between countries. With the advances in prenatal screening methods and with improved resolution and expertise at ultrasound scanning the questions of which screening test to use and when to offer ultrasound scans in pregnancy are difficult ones to answer.

In a previous report from 2004 we described the prenatal diagnosis policies in European countries. In this report we have updated the information with the policies in place in 2009. One EUROCAT representative from each country has written a chapter describing their national policy for prenatal screening under the four headings:

Screening for Down syndrome

Indications for prenatal cytogenetic diagnosis

Screening for Structural Anomalies by Ultrasound Screening

Termination of Pregnancy for Fetal Anomaly

Following the country chapters a list of recent EUROCAT publications in the area of prenatal diagnosis are given.

AUSTRIA

Written by Martin Haeusler (October 2009)

Screening for Down Syndrome

A national screening programme for congenital anomalies or aneuploidies is strictly avoided, because doctors and the national health service are NOT interested in terminating as many handicapped individuals as possible. But pregnant women have to be informed in time by the obstetricians about the existence of all the prenatal tests available, mentioning the advantages, the related risks and possible difficult decisions arising from these tests. An informed decision from the woman is sought.

Public insurance in Austria pays for CVS (chorionic villous sampling) or Amniocentesis if the pregnant woman is 35 years or older on EDD (estimated date of delivery). Furthermore the costs of Combined Tests and detailed ultrasound (20 weeks scan) are paid by public insurance since Sept 2009, if the pregnant woman is 36 years of age or older at conception. Further indications for offering these tests for free are: previous child with congenital anomaly or aneuploidy, consanguinity, suspicion of teratogenicity. All these non-invasive / risk calculation tests and invasive / diagnostic tests are offered on request of the pregnant women and are not compulsory.

Since Combined Tests are increasingly requested, the number of invasive tests is significantly declining.

Indication for Prenatal Cytogenetic Diagnosis

The invasive test has to be indicated and wanted by the relevant pregnant woman.

- Pregnant women 35 years or older on EDD
- Genetic disorders in parents and relatives
- Previous child with genetic disorder or metabolic defect
- Signs of developmental disorder in previous ultrasound examination
- Serological suspicion or sign of aneuploidy
- Intake of teratogens or high-dose radiation
- Extreme anxiety of pregnant women

Screening for Structural Anomalies by Ultrasound Scanning

All ultrasound examinations are voluntarily and not compulsory (see above).

The “Mother-Child-Passport” offers two sonographic examinations: at 20 and 30 weeks’ gestation. In the near future a third examination at 12 weeks’ gestation will be added.

The 20 weeks scan is targeting structural anomalies and markers for aneuploidy.

The National Ultrasound Society has published standards for these detailed scans. It is defined and published nationally, what has to be seen, how it has to be documented, and minimal standards of the technical equipment used. These examinations are mainly offered by obstetricians. Standards for medical training in prenatal sonography, certification and audits are planned and in discussion. Up to now, specialist knowledge has to be accumulated individually and voluntarily.

Three different levels of expertise were firstly defined by Prof. Hansmann in Germany and implemented nationally in 1983. Austria followed shortly after. The 2009 version of the “Mother-Child-Passport” offers the two above mentioned ultrasound examinations, but explicitly states, that the medical standard requested is a level-1 basic one, and that the patient was informed about the possibility of a level 2 or 3 scan elsewhere, which has to be paid privately without special indication (see above).

Termination of Pregnancy for Fetal Anomaly

Austrian law (StGb § 97) allows TOP before the beginning of labour if:

1. This is the only way to avoid a severe danger for the life or severe danger for the physical or mental health of the pregnant woman, or
2. The mental or physical health of the fetus is heavily in danger, or
3. The pregnant girl was < 14 years at conception.

In daily practice, there are three time periods, where different indications for TOP are defined:

Within 3 months after conception a pregnancy can be terminated on request of the pregnant woman on any reason (in most cases for socioeconomic problems).

Before viability (i.e. < 24 weeks' gestation) TOP on request is done for fetal anomaly or aneuploidy of sufficient severity. Minor forms like hexadactyly or isolated clefts, etc. will not be terminated in Austria. In this time period an isolated trisomy 21 is accepted as medical indication for a requested of TOP.

After potential viability is reached after 23 weeks' gestation, TOP requests are accepted only in non-viable cases or late diagnosis of very severe congenital anomalies (e.g. hydranencephaly, schicencephaly, brain tumor). In these cases the decision has to be reviewed by a multidisciplinary local committee, and a feticide is done before labour induction.

BELGIUM

Written by Vera Nelen and Yves Jacquemyn (November 2009)

Screening for Down Syndrome

The current policy for prenatal screening for Down syndrome in Belgium includes:

- nuchal translucency between 11 and 13 weeks of gestation
- first trimester serum test at 8-15 weeks of gestation

These tests are offered and reimbursed for all women regardless of age.

Indications for Prenatal Cytogenetic Diagnosis

Cytogenetic diagnosis (with amniocentesis or chorion villus sampling) is offered to all women:

- 36 years and older at expected time of delivery
- With calculated risk higher than 1/250 on the basis of the above mentioned tests
- Who have a previous child with chromosomal abnormality
- Who have, or their partners have, a chromosomal abnormality (e.g. translocation)
- Have a family history (mother or father's family) of DNA abnormality or metabolic disorder
- Have a previous child with a congenital abnormality
- Have ultrasound scan abnormalities suggestive of a chromosomal abnormality

The costs are reimbursed

Screening for Structural Anomalies by Ultrasound Screening

In Belgium ultrasound scans are reimbursed one in every trimester of pregnancy. Routine scan for determination of gestational age is done in the first trimester. Routine scan for structural anomalies is done in the 2nd trimester.

Specialised ultrasound for structural anomalies is indicated when:

- Presence of known risk factors for structural anomalies in the foetus:
 - Previous child with structural anomaly, Diabetes Mellitus in mother, use of anti-epileptics or other drugs that are known for teratogenic affects.

- Indications of a structural anomaly in a routine ultrasound scan

Termination of Pregnancy for Foetal Anomaly

In Belgium termination of pregnancy for foetal anomaly is allowed until the 12th week of gestation on simple maternal request and can be performed after 1 week waiting and confirmation of the desire to terminate by the woman. Parents have to be informed about all the facts concerning their situation and decide whether to terminate the pregnancy in a controlled facility.

After 12 weeks of pregnancy termination is only possible for medical reasons including maternal disease and foetal disease not amenable to cure. The decision has to be supported by at least two physicians. All terminations of pregnancy have to be reported and registered at a central review authority.

CROATIA

Written by Ljubica Odak and Ingeborg Barisic December 2009

Screening for Down Syndrome

In Croatia we have two types of screening tests for Down syndrome – Maternal serum screening and US (ultrasound) examination that are used in combination.

Several screening strategies are currently in use as there are no official guidelines.

Biochemical tests are available in major Croatian cities. Hospitals, private clinics and private laboratories offer different types of screening - double, triple, or combined test. Biochemical screening is offered to all pregnant women and costs are covered by national health insurance

Ultrasound first trimester screening is also offered to all pregnant women, alone or in combination with biochemical screening. US estimate of gestational age is done to improve the performance of biochemical tests. Ultrasonographic "soft marker" of Down syndrome and other chromosomal abnormalities are evaluated (nuchal translucency, cardiac echogenicities, choroids plexus cysts, renal calyceal dilation, echogenic bowel absence of nasal bone etc)

Costs of US screening in private institutions are not covered by basic health insurance. Women found to be at increased risk of having a baby with Down syndrome with first-trimester screening are offered genetic counselling and the option of CVS or mid-trimester amniocentesis.

Indications for Prenatal Cytogenetic Diagnosis

There are no official recommendations for prenatal cytogenetic diagnosis, but the following indications are widely used:

- Advanced maternal age (35 and over)
- Previous child (livebirth or stillbirth) with a chromosomal anomaly
- Parental chromosome rearrangement
- Abnormalities discovered during pregnancy: increased risk at maternal serum screening, abnormal foetal ultrasound etc.
- Exposure to radiation/chemotherapy

- Sex determination in X-linked disorders
- Paternal age above 42, and maternal anxiety are occasionally considered

Genetic counselling before and after cytogenetic investigation is required. Costs for cytogenetic prenatal diagnosis are covered by national health insurance.

Screening for Structural Anomalies by Ultrasound Scanning

There is no official recommendation, but usually three US examinations are performed during pregnancy. Screening for structural anomalies in:

- First trimester (10-12 weeks) includes measurements of foetal length, nuchal translucency, and nasal bone, determination of heart rate and evaluation of morphology of foetal brain, heart, limbs and abdominal wall.
- Second trimester, (18-24 gestational weeks) includes complete evaluation of the foetal morphology in order to detect congenital anomalies
- Third trimester, (28-32 gestational weeks) includes assessment of foetal growth

Termination of Pregnancy for Foetal Anomaly

Termination of pregnancy is regulated by the Croatian law in 1978 (NN 18/78). After the first 10 weeks of pregnancy, termination of pregnancy for foetal anomaly must be approved by a panel of experts, composed of two physicians, one of whom is gynaecologist, and a social worker or a registered nurse. The board may give consent to an abortion if congenital anomalies of foetus will result in a serious mental or physical handicap or if continuing pregnancy will endanger maternal health. Upper limit for termination of pregnancy for serious foetal anomaly in our country is 24-th gestational week.

DENMARK

Written by Marianne Christiansen (November 2009)

In 2004 a new national policy on prenatal diagnosis and screening was formulated in Denmark. Prenatal diagnosis and screening are free of charge for Danish citizens.

Screening for Down Syndrome

All pregnant women in Denmark are offered a first trimester risk assessment based on a first trimester serum test, the “Double test” (GA 8+0 to 13+6) and an ultrasound scan for Nuchal Translucency (GA 11+3 to 13+6).

Indications for Prenatal Cytogenetic Diagnosis

The indications for chorionic villus sampling or amniocentesis are:

- Women who have a calculated risk $\geq 1:300$ for Down Syndrome (on the time of screening), based on the combined first trimester test (serum test and Nuchal Translucency).
- Women who have previously given birth to a child with a chromosomal anomaly, or if one of the parents is a carrier of such.
- Women who have previously given birth to a child with a monogenetic disease, or if one of the parents is a carrier of such.
- If a close family member has a monogenetic disease.
- If an ultrasound scan has shown structural anomalies giving suspicion of a chromosomal anomaly.

Screening for Structural Anomalies by Ultrasound Scanning

All pregnant women are offered an ultrasound scan for structural anomalies at gestational week 19-20.

Termination of Pregnancy for Fetal Anomaly

The national policy (by law) is that up till 12 weeks of gestational age every woman may opt for a termination of pregnancy without special permission. After week 12 termination of pregnancy for fetal anomaly can be performed only after permission from a regional committee (two doctors and one employee at the Social Centre). The upper gestation limit for

termination of pregnancy for a fetal anomaly is usually week 22, unless it is a lethal anomaly. There should be no regional variations, and it is possible to appeal the decision.

FINLAND

Written by Annukka Ritvanen (February 2010)

Prenatal screening programmes have been harmonised in Finland from the beginning of the year 2010. The provision of screening was regulated by a Governmental Decree on Screening from 2006 (1339/2006, updated in 280/2009), which aims to increase the uniformity and quality of screening programmes. The municipalities and joint municipal boards have to organise the screening of foetal chromosomal anomalies and severe structural anomalies as a part of the general health care. The screening has to be offered for all pregnant women free of charge. The pregnant women can choose which of the voluntary prenatal screening programmes they want to participate, if any.

The national screening programme also includes guidelines on prenatal screening.

(<http://pre20090115.stm.fi/pr1178537602112/passthru.pdf>; updating of decree:

[http://finohta.stakes.fi/NR/rdonlyres/BACC269F-1987-4157-82AC-](http://finohta.stakes.fi/NR/rdonlyres/BACC269F-1987-4157-82AC-45B780560783/0/VN_asetus_seulonnoista_010509_alk.pdf)

[45B780560783/0/VN_asetus_seulonnoista_010509_alk.pdf](http://finohta.stakes.fi/NR/rdonlyres/BACC269F-1987-4157-82AC-45B780560783/0/VN_asetus_seulonnoista_010509_alk.pdf); tests:

http://www.stm.fi/c/document_library/get_file?folderId=39503&name=DLFE-6503.pdf).

Two information leaflets have been drawn on prenatal screening that can be handed out to expectant parents. One leaflet provides information on the available voluntary screening tests for chromosomal and structural anomalies. The other is intended for pregnant women who have received prenatal screening results with a suspected foetal chromosomal or structural anomaly. (<http://finohta.stakes.fi/FI/sikioseulonnat/perheille/index2.htm>) (also available in Swedish and English).

Screening for Down Syndrome

A general ultrasound screening during early pregnancy is offered for all pregnant women in gestational weeks 10+0–13+6. This screening aims at checking gestational age, plurality and place of placenta, not at detecting foetal anomalies although they may be found. If a pregnant woman has decided to participate in the prenatal chromosomal screening, primarily the early pregnancy combined screening is offered. A serum test during early pregnancy (S-PAPP-A and S-hCGB-V) is performed in gestational weeks 9+0–11+6 and measurement of the nuchal translucency combined with the general ultrasound screening during early pregnancy is performed in gestational weeks 11+0–13+6.

Alternatively, for instance when the first visit in the Maternity Health Care Centre takes place after week 11+6 and or when a reliable measurement of the nuchal translucency is not possible, a (two-marker) serum test (S-AFP, S-hCGB-V) of the second trimester is performed in weeks 15+0–16+6.

Both chromosomal screening options cannot be chosen. The purpose of both options is to find the pregnant women who have a higher risk for trisomies (21, 18, 13) and then to offer them a possibility for further chromosomal tests, chorionic villous biopsy (CVS) or amniocentesis (AC).

The municipalities have an option to offer direct chorionic villous sampling or amniocentesis for pregnant women aged 40 years or more.

Screening for Structural Anomalies by Ultrasound Screening

In order to detect severe structural defects a morphological ultrasound screening is performed primarily in gestational weeks 18+0–21+6 or alternatively after week 24+0. Both are not performed.

The primary purpose of the morphological screening in weeks 18+0–21+6 is to detect severe foetal structural anomalies so that further investigations can be performed before the week 24+1. The purpose of the alternative morphological screening (after week 24+0) is to detect foetal anomalies where the prognosis of the newborn can be improved by following up the pregnancy and giving birth at an optimal hospital, where both the pregnant mother and the newborn can get the necessary treatment without delay. Induced terminations of pregnancy due to foetal birth defects are not allowed after week 24+0.

In all, two prenatal ultrasound screenings are usually performed during pregnancy.

In accordance with the quality recommendations for prenatal screening, given by a Group of Experts in support of implementation of the Government Decree on Screenings (Ministry of Social Affairs and Health), general ultrasound screening during early pregnancy, early pregnancy combined screening and structural ultrasound screenings can be organised in either primary health care or in specialised medical care or partly in both. The laboratory examinations and risk calculation being part of the early pregnancy combined screening and

the serum screening during the second trimester of pregnancy should be centralised in university hospitals and other laboratories that are sufficiently large. Further (diagnostic) examinations related to screening should be centralised in university hospitals. The Expert Group considered that organising screenings within public health care will guarantee the best result. Municipalities may, however, purchase the prenatal screening from an outside service provider.

The ways of carrying out the municipal screening, the costs of screening and the quality monitoring system should be assessed in a few years. A national register for prenatal screening is being planned.

Indications for Prenatal Cytogenetic Diagnosis

In Finland the indications for prenatal cytogenetic diagnosis are:

- A higher calculated risk for Down's syndrome / chromosomal defects in prenatal screening tests,
- Maternal age of 40 years or more (optional in some municipalities),
- Previous child or foetus with a chromosomal anomaly or a monogenetic disease, or one of the parents a carrier for such, or
- Foetal major structural anomalies or soft markers or their combinations detected by prenatal ultrasound giving suspicion of a chromosomal or monogenetic defect.

Maternal wish for foetal chromosomal testing without other indications is only possible in private clinics and at own expense.

Termination of Pregnancy for Foetal Anomaly

According to the Finnish Abortion Law a pregnancy can be terminated for foetal indications (severe disease or birth defect) only on the permission from a national Abortion Committee in the National Supervisory Authority for Welfare and Health (Valvira) under the Ministry of Social Affairs and Health. A pregnancy can be terminated up to the end of 20th gestational week ($\leq 20+0$), when it is suspected that the child to be born might be mentally retarded or that the child might have or develop a severe disease or birth defect (risk for these). Valvira can also give permission for a termination after the 20th gestational week up to the end of 24th gestational week ($20+1-24+0$), if a severe foetal disease or birth defect has been

detected by a reliable prenatal test. After the end of 24th gestational week (> 24+1) termination of pregnancy for foetal indications is illegal. In practice, most severe foetal defects have been detected by a reliable prenatal test also in terminations of pregnancy for foetal indications performed before the 20+1 gestational week.

All terminations of pregnancy have to be reported and registered in the National Abortion Register and all foetal birth defects and diseases detected in terminations of pregnancy in the Finnish Register of Congenital Malformations, both maintained by the National Institute of Welfare and Health.

Further information on prenatal screening in Finland is to be found in <http://finohta.stakes.fi/FI/sikioseulonnat/index.htm>.

FRANCE

Written by Catherine de Vigan (December 2004). Updated by Babak Khoshnood in January 2010

Screening for Down Syndrome

The current policy for prenatal screening of Down's syndrome in France includes:

- Nuchal translucency measurement as a matter of routine between 11 and 13 weeks of gestation
- Maternal serum screening between 14 and 16 weeks, which should be systematically proposed to all women as stated by a law implemented in January 1997.

Costs of antenatal screening are reimbursed, and in the case of an abnormal result in any of the screening tests, amniocentesis is proposed and its costs are reimbursed.

Added 2010: Forthcoming changes in prenatal testing policy for Down syndrome: The French High Authority for Health (Haute Autorité de Santé, HAS) recently recommended the establishment of combined first-trimester screening with nuchal translucency measurement and first-trimester serum screening (free beta subunit of hCG and pregnancy-associated plasma protein A) for all pregnant women. The HAS also recommended that the implementation of this strategy should be accompanied by the establishment of a regulated quality-assurance program for the nuchal translucency measurement and serum screening tests in the first trimester. The HAS further asserted that the offer of invasive diagnostic testing to all women aged 38 years and older without prior screening is no longer justified (http://www.has-sante.fr/portail/jcms/c_540874/evaluation-des-strategies-de-depistage-de-la-trisomie-21). These recommendations for prenatal screening are likely to have major implications for the practice of Down syndrome screening in France.

Indications for Prenatal Cytogenetic Diagnosis

The policy of maternal serum screening implemented in 1997 and the new combined test screening includes offer of amniocentesis when the calculated risk is greater than 1/250.

In recent years, prenatal diagnosis of Down's syndrome has expanded considerably from a system based on offering amniocentesis (or chorionic villus sampling) to women 38 years of

age or older and those with a significant family history (paternal or maternal translocation, previous sibling with Down's syndrome) to a regulated system of universal access to screening.

In 1988, the indications for prenatal cytogenetic diagnosis were extended to include ultrasound abnormalities suggestive of a chromosomal anomaly. The indications for a cytogenetic diagnosis have been further extended to include "scan markers", including nuchal thickness greater than 3mm.

For all of the indications noted above, the costs of amniocentesis (or CVS) are fully reimbursed.

In addition to access to modalities of prenatal screening as described above, mothers 38 years of age or older have the option of direct access to reimbursed amniocentesis (i.e. without prenatal screening tests).

Screening for Structural Anomalies by Ultrasound Screening

Three routine scans for detecting structural abnormalities are performed during pregnancy. These examinations are performed at around 12 weeks, 22 weeks (morphological scan with cardiac examination) and 32 weeks of gestational age.

Terminations of Pregnancy for Fetal Anomaly

In the event of a prenatal diagnosis of Down's syndrome, or in general any "serious illness, recognized as incurable at the time of diagnosis", termination of pregnancy is allowed regardless of gestational age. Decisions for pregnancy terminations are reviewed by multidisciplinary committees for prenatal diagnosis.

IRELAND

Written by Bob Mc Donnell (December 2009)

Screening for Downs Syndrome

There is no policy for prenatal screening of Down Syndrome. Nuchal translucency measurement is not routinely offered, it is available if requested on an individual basis.

Indications for Prenatal Cytogenetic Diagnosis

Pre-natal cytogenetic diagnosis (amniocentesis or chorionic villus sampling) is not routinely offered, it is available if requested on an individual basis.

Screening for Structural Anomalies by Ultrasound Screening

A gestational age scan at approximately 18 weeks generally includes screening for structural anomalies.

Termination of Pregnancy for Fetal Anomaly

Termination of pregnancy is not legal for fetal anomaly.

ITALY

Written by Ettore Cariati, Francesca Strigini, Fabrizio Bianchi, Anna Pierini,

February 2010

Screening for Down Syndrome

The current policy for prenatal screening of Down's syndrome in Italy includes:

- Advanced maternal age (35 years or more at time of delivery)
- Maternal serum screening between 16th and 17th weeks by triple test (AFP, hCG, uE₃)
- Nuchal translucency measurement by ultrasound between 11th and 14th week, alone or combined with the maternal serum measurement of *free-β*-hCG and pregnancy associated plasma protein – A (double-test)

In some regional areas, like Tuscany, specific programmes for prenatal screening of Down's syndrome were developed, according to the guidelines of the *Fetal Medicine Foundation*.

In some private ultrasound centres, the sonographers perform the “genetic” sonogram, between 16th and 22nd week, but this procedure is not included in the public screening programmes.

Indication for Prenatal Cytogenetic Diagnosis

The cytogenetic diagnosis is offered free of charge to women:

- Aged 35 years or more at time of delivery
- Who have, or their parents have, a chromosomal abnormality
- Who have DNA abnormality or metabolic disorders in their or their spouse's family
- Who have a previous child with chromosomal abnormality
- Evidence of structural anomaly in fetal ultrasound scan
- Who have a 1st trimester nuchal translucency, or a “double test”, or a 2nd trimester triple test results 1:350 or higher (at time of delivery).

Screening for Structural Anomalies by Ultrasound Screening

In Italy there is a policy for three routine ultrasound examinations during the pregnancy:

- First trimester ultrasound screening, between 10th and 12th week. Purposes:
 - Pregnancy seat
 - Viability and number of fetuses

Biometric measurements (CRL, BPD) for dating of pregnancy

- Second trimester ultrasound screening, between 19th and 21st. Purposes:

Viability of the fetus

Biometric measurements (BPD, HC, AC, FL, TCD) for evaluation of fetal development

Evaluation of placenta location and amniotic fluid

Screening of fetal malformations

- Third trimester ultrasound screening, between 30th and 34th week. Purposes:

Viability of the fetus

Biometric measurements (BPD, HC, AC, FL) for evaluation of fetal development

Evaluation of placenta location and structure

Evaluation of amniotic fluid

Termination of Pregnancy for Foetal Anomaly

The national policy by law (n.194/1978) provides for the possibility of free abortion up to 12 weeks and 6 days of pregnancy. The only legal restriction is on the obligation to perform abortions in public hospitals.

After this time, the indication for the termination of the pregnancy of a malformed foetus is based on maternal physical or psychological disease, even if caused by awareness of a malformed foetus.

The reference time frame for legal limit of pregnancy termination is derived from evidence of survival possibility of foetuses which is generally not under 23 weeks of gestation. As it is difficult to evaluate this limit, lack of homogeneity among Regions may emerge.

MALTA

Written by Miriam Gatt (January 2010)

Screening for Down Syndrome

There is no policy for screening of Down Syndrome. Nuchal translucency not routinely performed.

Indications for Prenatal Cytogenetic Diagnosis

Pre-natal cytogenetic diagnosis (amniocentesis or chorionic villus sampling) is not performed.

Screening for Structural Anomalies by Ultrasound Screening

A gestational age scan at approximately 18 – 20 weeks gestation is routinely performed and this includes screening for gross structural anomalies.

Termination of Pregnancy for Fetal Anomaly

Termination of pregnancy is not legal for any reason including fetal anomaly.

THE NETHERLANDS

Written by Hermien de Walle (December 2009)

Screening for Down Syndrome

Every pregnant woman receives counseling in early pregnancy about screening for fetal anomalies. This counseling is reimbursed. She can choose to have:

- Nuchal translucency between 11 and 14 weeks of gestation
- Early serum analysis (PAPP-A and free beta hCG) between 9 and 14 weeks of gestation

When both tests are performed, the above is referred to as the 'combined test'.

In particular cases it is possible to have late serum analysis (serum- α FP, hCG and oestriol) at 15-19 weeks of gestation. This test is known as 'triple test'.

The costs of these tests are only reimbursed if the woman is 36 years of age or has an increased risk for fetal anomalies. The 'combination test' costs 140,- euro.

Indications for Prenatal Cytogenetic Diagnosis

Cytogenetic diagnosis (with amniocentesis or chorion villus sampling) is offered to all pregnant women:

- 36 years and older in the 18th week of pregnancy
- That have, or their partners have, a chromosomal abnormality (eg. translocation)
- Have ultrasound scan anomalies suggestive for a chromosomal abnormality
- Who had a previous pregnancy/child with chromosomal abnormality
- With additional risk to have a child with: an autosomal recessive disorder, autosomal dominant disorder or a X-chromosomal disorder
- With a detectable mitochondrial hereditary disease
- With a risk higher than 1/200 from screening (see above)
- Who became pregnant by an ICSI- procedure
- Who became pregnant by PGD-procedure

These costs are reimbursed.

Screening for Structural Anomalies by Ultrasound Screening

Every pregnant woman is offered a routine ultrasound scan at approximately 20 weeks of gestation. The aim of this scan is to check the development of the organs, growth of the fetus and the quantity of amniotic fluid. This routine scan can be performed by health professionals with ultrasound education e.g. midwives, general practitioners. The costs are reimbursed.

Indication for 'advanced ultrasound screening' around 20 weeks is the presence of known risk factors for structural anomalies in the fetus:

- Previous child or family member with structural anomaly, diabetes mellitus in mother, use of anti-epileptics or other drugs that are known for teratogenic effects.
- Exposure to X-radiation and/or chemotherapy
- Monochorial twin pregnancy
- Positive TSH-receptor antibodies
- Consanguinity
- The pregnant woman is 36 years or older and waived any screening or diagnostics in early pregnancy
- Indications for a structural anomaly or other significant findings in a routine ultrasound scan
- Growth deviations
- Oligohydramnion/polyhydramnion
- Fetal heart rhythm disorder
- Maternal infection that has possible effect on the fetus

Advanced Ultrasound Screening is only performed in a Center for Prenatal Diagnostics, by specialized gynaecologists. The costs are reimbursed.

Termination of Pregnancy for Fetal Anomaly

In the Netherlands termination of pregnancy is allowed until the 24th week of pregnancy, 24 weeks to be considered the limit of non-viability. After 24 weeks gestation termination of pregnancy is possible in only very limited cases. There are two categories:

- When the fetus has a disorder not compatible with life and the pregnant woman has fierce mental problems with carrying out the pregnancy. This is not punishable by law and does not need to be reported to the public prosecutor.
- The fetus could live after being born but postnatal life-prolonging medical treatment is considered to be no useful purpose. This has to be reported to the public prosecutor, who will decide whether or not it is a punishable act in this particular case.

SPAIN

(Barcelona/Catalunia)

Written by Joaquín Salvador (January 2010)

Screening for Down Syndrome

The current policy (from October 2009) for prenatal screening of Down syndrome in Catalonia includes:

To all women: First trimester screening:

- Routine nuchal translucency measurement between 11 and 13 weeks of gestation.
- Routine maternal serum screening (free β hCG and PAPP-A) between 8 and 13 weeks of gestation.

To women without first trimester obstetrics visits: Second trimester screening:

- Maternal serum screening (free β hCG, α FP, uE3 and inhibin A) from 14 weeks of gestation.

Indications for Prenatal Cytogenetic Diagnosis

- Calculated risk of ≥ 250
- Mother or father carrier of balanced translocation.
- Previous pregnancy with chromosomal anomaly.
- Family history of genetic disorder with available prenatal diagnostic.
- Birth defect detected by obstetric ultrasound (this indication is not well defined).

Screening for Structural Anomalies by Ultrasound Screening

Three routine scans (1/trimester of pregnancy).

Terminations of Pregnancy for Fetal Anomaly

Induced abortion after prenatal detection of fetal anomaly is legal in Spain until 22 weeks of gestation. A new law has been approved by the Spanish Parliament that allows pregnant women to induce for abortion fetuses with major birth defects detected after 22 weeks of gestation.

All these procedures are theoretically free of charge (included in the Public and Universal Spanish Health System). In practice, in Barcelona some percentage of terminations are performed at private clinics.

SWEDEN

Written by Göran Annerén (January 2010)

Screening for Down Syndrome

No policy for prenatal screening of Down syndrome exist in Sweden. Nuchal translucency measurement and chemical blood test as a matter of routine between 11 and 13 weeks of gestation has been introduced in some parts of Sweden

Indications for Prenatal Cytogenetic Diagnosis

Prenatal diagnosis of Down syndrome is based on offering amniocentesis(or chorionic villus sampling) to women 35 years of age or older and those with a significant family history (paternal or maternal translocation, previous sibling with Down syndrome) and to those with abnormal screening results to a regulated system of universal access to screening. Half of all pregnant women, who are offered prenatal diagnosis, will today use this offer. If the woman is younger than 35 years of age, but very worried of having a child with Down syndrome, she is offered prenatal diagnosis. The costs of screening, amniocentesis (or CVS) are fully reimbursed.

Screening for Structural Anomalies by Ultrasound Screening

One routine scan for detecting structural abnormalities is performed during pregnancy. These examinations are performed at around 15-18 weeks (morphological scan with cardiac examination)

Terminations of Pregnancy for Fetal Anomaly

In the event of a prenatal diagnosis of Down syndrome, or in general any "serious illness, recognized as incurable at the time of diagnosis", termination of pregnancy is allowed before the end of 18 weeks gestation. For pregnancies that are >18 weeks gestation decisions for pregnancy terminations are reviewed by multidisciplinary committee at the National Board of Health and Welfare. Few terminations of pregnancy are permitted after 22 weeks gestation.

SWITZERLAND

Written by Marie-Claude Addor in January 2005 and reviewed in December 2009. No changes in policies over this 5 year period.

Screening for Down Syndrome

Screening for Down Syndrome should be offered to every woman independently of maternal age. Gynecologists are encouraged to perform the first trimester maternal blood test rather than the 2nd trimester because of its better accuracy. The 1st trimester screening is performed between the 10th and the 14th week of gestation. It is based on the measurement of the nuchal translucency by ultrasound combined with the maternal age and the levels of pregnancy associated plasma protein A (PAPP-A) and free subunit of beta human chorionic gonadotrophin (β -hCG). The 2nd trimester screening is performed between the 15th and the 19th week of gestation. It is based on the levels of alpha-fetoprotein (AFP), β -HCG and unconjugated estriol (uE3) of the maternal serum. In the cases where a 1st trimester screening is chosen, a 2nd trimester assay of AFP is recommended to exclude open neural tube defects.

Indication for Prenatal Cytogenetic Diagnosis

Prenatal cytogenetic diagnosis is offered to every woman whose 1st trimester screening established a risk over 1/300 or 2nd trimester screening showed a risk over 1/380.

Prenatal cytogenetic diagnosis is also offered to women who are over 35 years old and to patients where ultrasound scanning suggested an anomaly. It can also be offered when the couple wishes to undertake such test. If the test is performed only for parental anxiety, it is billed to the couple.

Women are referred by their gynecologists either directly to the ultrasound service where the amniocentesis or CVS is performed or to the genetic service where they have counseling beforehand. In the latter situation, a medical geneticist or a genetic counselor sees the patient half an hour before the amniocentesis or CVS. During the counseling session, the professionals gather information about the pregnancy, draw the pedigree, discuss the screening results, explain the technique, discuss its risks and other available options (screening for women who were referred for maternal age only). They also answer the patients' questions and listen to their anxieties.

Screening for Structural Anomalies by Ultrasound Scanning

The first ultrasound scanning is performed between the 10th and the 12th week of the pregnancy and the second between the 20th and the 23rd week. When risk factors are present, extra tests may be performed. The 1st ultrasound scan helps to determine the gestational age of the fetus and measures of the nuchal translucency. These data are used in the 1st and 2nd trimester maternal serum screening. The 2nd ultrasound scan assesses for fetal malformations, growth delay and amniotic fluid quantity.

Termination of Pregnancy for Fetal Anomaly

According to the Swiss penal code there is no legal limit for termination of a pregnancy according to gestation age. However in practice, it is performed until the 24th week of gestation.

UK

Written by Patricia Boyd (December 2009)

The UK National Screening Policy position is that screening for Down's syndrome should be offered to all pregnant women and that all trusts must ensure that they provide a dating scan, and 18-21 week anomaly ultrasound scan, in line with NICE (National Institute for Health and Clinical Excellence) and UK National Screening Committee recommendations.

Screening for Down Syndrome

England - The UK National Screening Committee (UK NSC) recommends the following outcomes and benchmark time frames for the Down's syndrome screening programme in England:

Programme Outcomes

- A detection rate (DR) for Down's syndrome of greater than 75% of affected pregnancies with a screen positive rate (SPR) of less than 3%. (*Benchmark time frame: April 2007 to April 2010*)
- A detection rate (DR) of greater than 90% of affected pregnancies with a screen positive rate (SPR) of less than 2%. (*Benchmark time frame: by April 2010*)

In practice the type of test offered varies throughout England with most of the Southern half offering first trimester Combined testing and the Northern half offering second trimester Triple or Quadruple tests.

Wales - Standards for Down's syndrome screening and ultrasound scanning are set by Antenatal Screening Wales, which is part of Public Health Wales.

The National Assembly for Wales policy is that:

- All women should be offered screening for Down's syndrome which provides a minimum of a 60% detection rate for a 5% false positive rate and is supported by early pregnancy ultrasound scanning.

Currently all women are offered the second trimester Down's syndrome Triple screening test.

Scotland - All pregnant women in Scotland are currently offered a second trimester serum screening test which includes the measurement of 2 biochemical markers in the second trimester of pregnancy. Combined first trimester screening is being introduced and will enable the programme to meet the NSC's current standard which requires the programme to achieve a 75% detection rate with less than 3% false positive rate.

Northern Ireland - No official policy and no screening except by private tests.

Indications for Prenatal Cytogenetic Diagnosis, England, Wales and Scotland

Maternal age (usually over 35 years), high risk Down's syndrome screening test result, family history of chromosome anomaly, translocation carrier, and ultrasound malformations are all indications for prenatal cytogenetic diagnosis.

With the move from maternal age screening to serum screening and nuchal translucency scanning the numbers based on maternal age alone are diminishing.

Screening for Structural Anomalies by Ultrasound Screening

The National Institute for Clinical Excellence, the NHS Fetal Anomaly Screening Programme (England), Antenatal Screening Wales and the Scottish Executive all recommend that every pregnant woman should be offered a fetal anomaly ultrasound scan between 18 weeks, 0 days and 20 weeks, 6 days.

In Northern Ireland all Trusts offer all pregnant women an ultrasound scan for fetal abnormalities in the second trimester of pregnancy.

Termination of Pregnancy for Fetal Anomaly (TOPFA) - England, Wales and Scotland

The law allows TOPFA if the pregnancy "has NOT exceeded its 24th week and the continuance of the pregnancy would involve risk, greater than if the pregnancy were terminated, or injury to the physical or mental health of the pregnant woman".

There is no gestation limit if "There is substantial risk that if the child were born it would suffer from such physical or mental abnormalities as to be seriously handicapped".

There is also no gestation limit if “the continuance of the pregnancy would involve risk to the life of the pregnant woman greater than if the pregnancy were terminated”.

In practice there is regional variation in availability of late TOPFA.

In Northern Ireland termination of pregnancy for fetal anomaly is illegal.

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