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## **RESEARCH ARTICLE**

## PRENATAL DIAGNOSIS ANDCOUNSELLING

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## **ARTICLE INFO**

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## ABSTRACT

Preconception health care is the medical care a person receives from heath team members for better outcome in pregnancy. The objective of prenatal diagnosis and counselling is to reduce complications by identifying the risk factors early. Through proper prenatal diagnosis helps to reduce maternal and fetal mortality and morbidity rate. Prenatal diagnosis helps the couple to take prompt treatment to avoid complications in pregnancy. Couples have to be educate about the importance of antenatal visit and different measures for identify risk factors. Preconception care can significantly improve maternal and infant outcomes, and thus optimise intergenerational health. The aims of this scoping review are to (1) provide an up-to-date summary of preconception health and care strategies, policies, guidelines, frameworks and recommendations across the UK and Ireland and (2) explore preconception health and care services and interventions in Northern Ireland as a case study<sup>1</sup>Preconception health and health care focus on things you can do before and between pregnancies to increase the chances of having a healthy baby. For some people, getting their bodies ready for pregnancy takes a few months.2. Some parents have increased risk of having a baby with a genetic disorder or other problem. They may want to consider one of these tests. Knowing about problems before the baby is born may help parents. They may be able to make better decisions about health care for their infant. Certain problems can be treated before the baby is born. Other problems may need special treatment right after delivery. In some cases, parents may even decide not to continue the pregnancy.<sup>4</sup>The main benefit of CVS is it can be done earlier in the pregnancy. It's very accurate in detecting genetic abnormalities. But it does not detect some things that amniocentesis like Neural tube defects Birth defects incompatibility Amniocentesis might be the best option if previous history of a neural tube defect. The results of other tests during your pregnancy have been abnormal.

## **INTRODUCTION**

Prenatal diagnosis stress the need to detect early in pregnancy a number of foetal anomalies and genetic diseases. The prenatal diagnosis of genetic diseases has become widely available for pregnancies at risk in the last three decades Prenatal diagnosis allows couples at risk to envisage a pregnancy since an alternative is now offered to them. Congenital abnormalities account for 20-25% of perinatal deaths. Now, many genetic and other disorders can be diagnosed early in pregnancy.<sup>3</sup>

#### Prerequisites

- The pregnant woman is 35 years or older at the time of delivery.
- She or her parents have had a previous child with a chromosomal abnormality.

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- She has a history of recurrent abortions, or her husband's previous wife experienced several miscarriages.
- A history of parental consanguinity is present.
- The couple is known to be carriers of a chromosomal translocation.
- The pregnant woman is affected with type 1 diabetes mellitus, epilepsy, or myotonic dystrophy.
- She is exposed to viral infections, such as rubella or cytomegalovirus.
- The mother is exposed to excessive medication or to environmental hazards.
- In her or her spouse's family, a history of Down syndrome or some other chromosomal abnormality is present.
- A history of single gene disorder is present in her or her spouse's family.
- Her male relatives have Duchenne muscular dystrophy or severe hemophilia.
- She is suspected of having some other harmful gene on her X chromosomes.
- The fetus is diagnosed in utero to have some hereditary error of metabolism.
- The fetus is detected to be at increased risk for a NTD<sup>3</sup>

## **BENEFITS OF PRENATAL DIAGNOSIS**

- Prenatal diagnosis determines the outcome of pregnancy.
- It is helpful for couples to decide whether to continue the pregnancy.
- It indicates possible complications that can arise at birth process.
- Prenatal diagnosis is helpful for the management of remaining weeks of pregnancy.
- It prepares the couple for the birth of a child with an abnormality.
- Prenatal diagnosis can be helpful for the improvement of the outcome of pregnancy using fetal treatment.

## **TYPES OF PRENATAL DIAGNOSIS<sup>8</sup>**

## NONINVASIVE TECHNIQUES

## Fetal visualization

- Ultrasound
- Fetal echocardiography
- Magnetic resonance imaging (MRI)
- Radiography

## Screening for neural tube defects (NTDs) - Measuring maternal serum alpha-fetoprotein (MSAFP)

## Screening for fetal Down syndrome

- Measuring MSAFP
- Measuring maternal unconjugated estriol
- Measuring maternal serum beta-human chorionic gonadotropin (HCG)

#### Separation of fetal cells from the mother's blood

Assessment of fetal-specific DNA methylation ratio

## **INVASIVE TECHNIQUES**

#### Fetal visualization

- Embryoscopy
- Fetoscopy

## Fetal tissue sampling

- Amniocentesis
- Chorionic villus sampling (CVS)
- Percutaneous umbilical blood sampling (PUBS)
- Percutaneous skin biopsy
- Other organ biopsies, including muscle and liver biopsy

## Preimplantation biopsy of blastocysts obtained by in vitro fertilization

## Cytogenetic investigations

- Detection of chromosomal aberrations
- Fluorescent in situ hybridization

## Molecular genetic techniques

- Linkage analysis using microsatellite markers
- Restriction fragment length polymorphisms (RFLPs)
- Single nucleotide polymorphisms (SNPs)
- DNA chip
- Dynamic allele-specific hybridization (DASH)

## NONINVASIVE TECHNIQUES<sup>8</sup>

## Fetal visualization

## Ultrasound

- No known risks to fetus or mother.
- Recommended for all pregnant women to determined gestational age, fetal viability, and multiple pregnancies.
- An integral part of amniocentesis, chorionic villus sampling and fetal blood sampling
- It provides detail visualization of foetus anatomy irrespective of its position or movement, assessment of amniotic fluid and localization of placenta.
- Helps to identifying some major malformations during 2nd or 3rd trimester Although ultrasonography provides only structural information but some structural abnormalities strongly suggest predisposition to genetic abnormalities or obstetric abnormalities and also help interpret abnormal maternal serum marker levels.

## **3D-ultrasound**

- Modern three-dimensional ultrasonography allows the direct assessment of fetal anomalies by providing surface-rendered and more transparent images. It is generally recommended for the couple having
- Family history of congenital malformations (pyloric stenosis, congenital heart defects, cleft lip and palate),
- Renal malformations (renal agenesis Potter's syndrome, polycystic kidney disease),
- Lethal forms of short-limbed skeletal dysplasias (thanatophoric skeletal dysplasia, achondrogenesis),
- Gut malformations (obstruction), diaphragmatic hernia, microcephalus, and hydrocephalus etc.

The major advantage of using 3D-ultrasound is that it supports the parents in making reproductive choices if fetus has severe anomaly however in negative case it can better assured the parent and the physician about the overall development of the fetus.

**Importance of accurate assessment of gestational age:** Accurate assessment of gestational age is particularly important in various intrauterine growth abnormalities associated with chromosomal disorders.

## Indication for Ultrasonography

- Previous child with known disorder such as Down syndrome
- To delineate malformation in fetus following biochemical or cytogenetic analysis
- In low risk pregnancy, however it requires greater expertise to screen for malformations

#### Advantages of using Ultrasonography are

- Low cost, easily accessible
- Accurate assessment of gestational age
- Detection of ectopic or non-viable pregnancies
- Assessment of fetal growth
- Detection of abnormal pregnancy using biometric parameter and heart rate
- Detection of initial fetal anomalies (anencephaly or abdominal wall defects)
- Identification of fetal anomalies or growth disturbances during the third trimester of pregnancy
- Detection of twin pregnancy
- Detection of amniotic fluid volume and its associated anomalies
- Locating position of placenta
- Monitoring movement of needle while performing invasive procedures

## Fetal echocardiography

Fetal echocardiography can be performed at 15 weeks' gestation and beyond. When this technique is used with duplex or color flow Doppler, it can identify a number of major structural cardiac defects and rhythm disturbances. Fetal echocardiography is recommended in cases where cardiac defects are suspected, including the following:

- Identification of an extracardiac malformation on routine ultrasound
- Abnormality of another major organ system
- Suspected genetic disease or fetal chromosome abnormality associated with heart defects
- Exposure to potentially teratogenic agents
- Family history of congenital heart defects, particularly in a parent or sibling
- Maternal diseases, such as diabetes or phenylketonuria associated with fetal structural heart defects, in particular heart blocks, such as lupus or other immune disorders
- Alcohol or drug consumption by mother during pregnancy
- Maternal rubella infection during pregnancy

#### MRI

MRI is a fetal imaging technique that uses powerful magnets and radio waves to construct images of the body, but, because of fetal movements, its application has been limited. Glenn et al reported a prenatal diagnosis of polymicrogyria using MRI as an imaging technique.

## Radiography

The fetal skeleton can be visualized by radiography from 10 weeks' gestation onward. This technique is used for the diagnosis of inherited skeletal dysplasias, particularly osteochondrodysplasia, in the second and third trimesters. Aslan et alcarried out prenatal diagnosis of thanatophoric dysplasia at 19 weeks' gestation in an 18-year-old woman. Because of the dangers of radiography to the fetus, this technique rarely is used.

## Screening for neural tube defects

# Screening for NTDs is recommended if the following are present:

- Ultrasound findings indicate NTDs.
- A child with NTDs is already in the family.
- A family history of NTDs exists, especially a mother with NTDs.
- The mother has type 1 diabetes mellitus during pregnancy.
- Maternal exposure to drugs, such as valproic acid, is associated with NTDs.
- Elevated level of MSAFP is present.

#### Measuring maternal serum alpha-fetoprotein

- Determine the AFP levels from the fetus. AFP is produced by the yolk sac and later by the liver; it enters the amniotic fluid and then the maternal serum via fetal urine.
- In open NTDand obstetrics emergencies (eg, anencephaly, spina bifida) and abdominal wall defects in the fetus, AFP diffuses rapidly from exposed fetal tissues into amniotic fluid, and the MSAFP level rises
- Performed between 15-22 weeks' gestation.
- MSAFP test and ultrasonography detects almost all cases of anencephaly and most cases of spina bifida.
- If the level of acetylcholinesterase rises along with AFAFP, it is suspected as a condition of a NTD.

#### Screening for fetal Down syndrome

- Measuring maternal serum alpha-fetoprotein
- Low level of MSAFP indicates Down syndrome or other chromosomal aneuploidy and failing pregnancies.
- Measuring maternal unconjugated estriol
- In the third trimester, the level of estriol gives an indication for the well-being of the fetus. A low level of estriol is an indication of Down syndrome and adrenal hyperplasia with anencephaly. If the estriol level drops to a great level, then it indicates risk to fetus.
- Measuring maternal serum beta-human chorionic gonadotropin

Beta-HCG, is an indication for pregnancy.

An increased beta-HCG level coupled with a decreased MSAFP level suggests Down syndrome. If it is lower than expected, it indicates abortion or ectopic pregnancy. If the level of HCG is considerably high, then it indicates the possibility of trophoblastic diseases. The elevated level of HCG, along with absence of the fetus on ultrasonography, indicates a hydatidiform mole.

## Measuring maternal inhibin-A levels

The hormone inhibin is secreted by the placenta and the corpus luteum. Inhibin-A can be measured in maternal serum.

An increased level of inhibin-A is linked with an increased risk for trisomy 21. A high inhibin-A level may also be associated with a risk for preterm delivery.

- Cell-free fetal nucleic acids from the placenta
- It can be used to screen for Down syndrome.
- Sex determination for families with inherited sex-linked diseases, diagnosis of certain single gene disorders, and blood Rhesus factor status (in the case of Rhesus D-negative mothers) can also be performed using cell-free fetal nucleic acids from the placenta.

**Separation of fetal cells from the mother's blood:** Fetal blood cells make access to maternal circulation through the placental villi. It can be collected safely from approximately 18 weeks' gestation onwardand collected at 12 weeks' gestation through successful procedures. Fluorescent in situ hybridization (FISH) is one technique that can be used to diagnose aneuploid conditions, such as trisomies and monosomy X. In the condition of fetal infection with such viruses as rubella, cytomegalovirus, and toxoplasmosis, the viral immunoglobulin M (IgM) or DNA also can be identified in fetal blood.

#### Fetal-specific differentially methylated regions

The free fetal DNA (ffDNA) is present in the maternal circulation during pregnancy. that analyzes fetal-specific differentially methylated regions (DMRs). Methylation ratio of normal and trisomy 21 cases for each tested fetal-specific DMR present in maternal peripheral blood and could diagnose 14 cases of trisomy 21 and 26 normal cases. Routine practice in diagnostic laboratories for all pregnancies.

## **INVASIVE TECHNIQUES<sup>8</sup>**

## Fetal visualization

**Embryoscopy:** Performed in the first trimester of pregnancy (up to 12 weeks' gestation). technique, A rigid endoscope is inserted via the cervix in the space between the amnion and the chorion, under sterile conditions and ultrasound guidance, to visualize the embryo for the diagnosis of structural malformations.

#### Fetoscopy

- Performed during the second trimester (after 16 weeks' gestation).
- A fine-caliber endoscope is inserted into the amniotic cavity through a small maternal abdincision, under sterile conditions and ultrasound guidance, for the visualization of the embryo to detect the presence of subtle structural abnormalities. it is also used for fetal blood and tissue sampling.



## Fetal tissue sampling

Amniocentesis (9,10)

## Amniocentesis is the sampling of amniotic fluid during pregnancy. It can be done:

- Early in pregnancy (early amniocentesis between 12 and 14 weeks' gestation).
- Later in pregnancy (midtrimester amniocentesis between 16 and 18 weeks' gestation).
- The fluid extracted contains cells from the amnion and fetal skin, lungs and urinary tract. The cells are grown in culture media allowing chromosomal, genetic, biochemical and molecular biological analysis:
- It is used to achieve prenatal diagnosis. Examples of diagnosis include:
  - For management of rhesus disease or estimation of maturity.
  - Chromosomal, genetic, biochemical and molecular biological analysis of cultured amniotic cells.
  - It is an invasive test posing risk to fetus and mother.
- It is not used as a screening test.

**Indications for amniocentesis:** Guidance from the National Institute for Health and Clinical Excellence gives some perspective to the place of amniocentesis in antenatal care. It is important to highlight that the list of indications will in practice be much shorter with better genetic counselling and the need for earlier diagnosis which often makes CVS a better option.

- Advanced maternal age (>35 years) the most common indication
- Previous child with:
  - Neural tube defect (1 in 20 subsequent pregnancies affected)
    - Chromosomal abnormalities
  - A birth defect
- Positive antenatal screening tests, including for example:
  - Fetal ultrasound findings
  - Raised maternal serum alpha-fetoprotein (ultrasound now also used in neural tube defect screening)
- History of:
  - Parent carrying a balanced chromosomal translocation (1 in 4–10 chance of a fetus being affected)
  - Risk of recessively inherited metabolic disorder
  - Mother carrying X-linked disorder(to determine fetal sex)
  - Mother exposed to certain drugs or infections (which can cause fetal malformations)
- Analysis to detect specific conditions from:
  - DNA (for example, fragile X syndrome, sickle cell disease, cystic fibrosis)
  - Enzymatic activity in amniocytes (for example, Tay-Sachs disease)
  - Fluid biochemistry (for example, in congenital adrenal hyperplasia - 17-OHprogesterone)

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• Note that testing for lung maturity is relevant only much later in pregnancy rather than in antenatal screening.

#### Procedure

#### Technique

- Rhesus immunoprophylaxis should be given where appropriate (fetomaternal transfusion is a risk in amniocentesis and chorionic villus sampling)
- Preferably under ultrasound guidance
- 22G spinal needle is inserted through the maternal abdominal and uterine walls into the pocket of amniotic fluid within the amniotic sac
- 10–20 ml of fluid is aspirated (or approximately 1 ml per week of gestation)
- A cell filtration system may be used
- Smaller volumes may be aspirated where advanced laboratory techniques require less material

#### • Midtrimester amniocentesis:

- Normally performed in the second trimester from 14–16 weeks' gestation
- There is relatively more amniotic fluid (enough amniotic fluid for reliable cell culture about 20 mls)
- There is still time to terminate the pregnancy (if results indicate this to be advisable)

## • Early amniocentesis:

- This has been conducted at weeks 9–14
- Less fluid is removed and ultrasound guidance is essential
- Carries a higher risk of loss of pregnancy (around 7%) and talipes equinovarus
- Preferred over chorionic villus sampling (CVS) where CVS unreliable (in twin pregnancies)

#### Amniocentesis

**Diagnostic testing of amniotic fluid:** It is useful to know what is offered locally as there is some variation across the country. The following tests can be performed:

#### On the amniotic fluid

- Alpha-fetoprotein (AFP) and acetylcholinesterase levels (for neural tube defects)
- Bilirubin levels (for gestational assessment and to detect isoimmune haemolysis)
- Tests of lung maturity (various but lecithin to sphingomyelin ratio for example)
- Enzyme analysis (many and varied including for inborn errors of metabolism)
- On fetal cells extracted from amniotic fluid testing for genetic and chromosomal disorders:
  - Rapid testing (results in 24 to 72 hours). In most areas this will identify specifically:
- Down's syndrome(trisomy 21)
- Edward's syndrome(trisomy 16)
- Patau's syndrome(trisomy 13)
- Turner's syndrome

- Klinefelter's syndrome
- Other sex chromosome anomalies (in some laboratories, not all)
  - Chromosome analysis after cell culture (results take about 2 weeks). This will give full karyotyping but even this will not identify all chromosomal abnormalities.
- Other possible tests on fetal cells (hardly used in practice with appropriate genetic counselling)
  - Direct DNA analysis techniques (for example for Tay-Sachs disease, phenylketonuria, Duchenne's muscular dystrophy and cystic fibrosis)
  - Indirect DNA analysis (used for example to detect linkage disorders when the exact gene is not known)
- Patients should be advised of how and when results of testing will be available and this may vary according to the tests being done and the laboratory used.
- Risks and complications of amniocentesis
- Distressing symptoms (uterine cramping)
- Uterine bleeding (about 2%)
- Amniotic fluid leakage (about 3%)
- There is a risk of maternal rhesus sensitisation in susceptible pregnancies (true for CVS as well)
- Amnionitis (only about 0.1%)
- 0.5 1% increased risk of pregnancy loss compared with the background risk<sup>7</sup>
- Failure of cell culture from 1% up to 5% if performed under 12 weeks' gestation
- Anxiety for parents, caused by delay in diagnosis (may make choices for termination of pregnancy difficult)

#### **Chorionic villus sampling**

**Indication for CVS:** Clinical referral for CVS is similar to those suggested for amniocentesis especially in the first trimester. In addition to karyotyping several other single gene disorders can be detected using chorionic villus sample. Examples are given below

- Fragile X syndrome
- Hemophilia A and B
- Haemoglobincpathy
- a thalassaemia
- Zellweger syndrome
- Tay-Sachs disease
- Cystic Fibrosis
- Duchenne muscular dystrophy
- Phenylketonuria (PKU)3
- Huntington's disease

**Procedure:** The chorionic villi sampling is performed by two different methods which are discuss below separately.

The transcervical method: The transcervical procedure is performed aseptically under ultrasound guidance. After assessing fetal viability a portex catheter is inserted into the cervix while keeping the patients in lithotomy position. A plastic cannula carrying obturator is then placed inside the placenta; once the catheter reaches its target obturator is replaced with the syringe containing medium and the chorionic villi is aspirated by moving the catheter to-and-fro under negative pressure. The transabdominal method: The Transabdominal CVS method can be performed at any stage of pregnancy following assessment of fetal heart rate and placental position by ultrasound. An 18-20 mm gauge needle is inserted transabdominally into the placenta and sample is withdrawn under continuous negative pressure using syringe carrying 2-3ml of transport medium. If the sample drawn is insufficient, then the procedure can be repeated at least twice however this would increase the rate of spontaneous abortion. The aspirated sample can be stored at room temperature still it is preferable to confirmed the conditions from the specialized laboratories.

**Cordocentesis/ Percutaneous umbilical blood sampling:** Cordocentesis also referred as fetal blood sampling process allows rapid detection of fetal abnormalities particularly in the second or third trimester of pregnancy. Since the test provides direct access to the fetal blood it helps monitor the pathological and physiological changes occurring in developing fetus. Additionally, it plays a major role in confirming fetal involvement of genetic mosaicism reported in 0.1-0.3% amniocentesis cases.

**Procedure:** The procedure is performed under the guidance of high-resolution ultrasonography by giving local anesthesia. After identifying position of placenta a stylet needle of 20-22 gauges is inserted into the abdomen. With lateral or posterior position of placenta, needle is pushed through the amnion to the placental insertion site to puncture umbilical cord. The blood will be collected using tuberculin syringe of I ml capacity. The most preferable puncture site is placental insertion site of umbilical cord or intra-abdominal umbilical vein; umbilical arteries are specifically avoided to prevent bleeding following puncture. Blood drawn from umbilical cord is directly used for fetal karyotyping, DNA based test, biochemical and serological assays. In addition, a sample of maternal blood is also drawn before the procedure for quality control against fetal blood sample.

**Post procedure monitoring:** After successful aspiration of fetal blood, punctured site should be monitor carefully for possible bleeding and if indicated fetal heart rate at least 1-2 hours. Since contamination with maternal blood or amniotic fluid may interfere with accurate diagnosis, purity of fetal blood specimen is confirmed using red blood size determination test (MCV); as fetal red blood cells are larger than maternal blood cells. Alternatively, sample can also be tested by blood typing (I antigen present only on maternal blood), determining the hCG concentration or Kliehauer-Betke test.

## Few important considerations before performing cordocentesis

- Positive history of chromosomal disorders such as polysploidy
- If required amniocentesis should be performed before cordocentesis to avoid contamination of amniotic fluid with fetal blood
- It is always recommended to use umbilical vein rather than an artery foi drawing the blood; this would help avoid longer post procedure bleeding and bradycardia.
- Sensitize Rh negative women by administering Rhlg.

#### Cordocentesis

#### Indications for fetal blood sampling

- Fetal hematologic disorders:
- Fetal infection:
- Acid-base balance in fetal growth restriction:
- Karyotyping:
- Suspected fetal thrombocytopenia
- Twin-Twin transfusion syndrome
- Fetal thyroid disorders
- Suspected fetal anemia
- Abnormal Doppler flow
- Duodenal atresia
- Single umbilical artery
- Omphalocele

#### Percutaneous skin biopsy

It performed under ultrasonic guidance between 17-20 weeks' gestation to prenatally diagnose a number of serious skin disorders, such as anhidrotic ectodermal dysplasia, epidermolysis bullosa letalis, epidermolysis bullosa dystrophica, hypohidrotic ectodermal dysplasia, oculocutaneous albinism, and genetic forms of ichthyosis,

**Other organ biopsies, including liver and muscle biopsy:** To diagnose an inborn error of metabolism, such as ornithine transcarboxylase deficiency glucose-6-phosphatase deficiency, glycogen storage disease type IA, nonketotic hyperglycemia, and carbamoyl-phosphate synthetase deficiency. It is best performed between 17-20 weeks' gestation under ultrasound guidance.

**Preimplantation biopsy of blastocysts obtained by in vitro fertilization:** Techniques are being developed to test cells obtained from biopsy of early cleavage stages or blastocysts of pregnancies conceived through in vitro fertilization. These techniques will be helpful for selective transfer and implantation of those pregnancies into the uterus that are not affected by a specific genetic disorder. This approach will be more acceptable to those couples who oppose abortions.

#### CYTOGENETIC INVESTIGATIONS<sup>7</sup>

#### **Detection of chromosomal aberrations**

- Chromosomal aberrations, such as deletions, duplications, translocations, and inversions diagnosed in affected parents or siblings, can be detected prenatally in a fetus by chromosomal analysis (see image below).
- Prenatal diagnosis for congenital malformations and genetic disorders. Karyotype showing trisomy 21 (47, XY, +21) in a male.
- This analysis can be undertaken on fetal cells obtained through such techniques as amniocentesis and CVS.

#### Fluorescent in situ hybridization<sup>7</sup>

• FISH uses different fluorescent-labeled probes, which are single-stranded DNA conjugated with fluorescent dyes and are specific to regions of individual chromosomes. These probes hybridize with complementary target DNA sequences in the genome and can detect chromosomal

abnormalities, such as trisomies, monosomies, and duplications.

• Three types of DNA probes are used in FISH analysis. Whole chromosome probes are specific to a whole chromosome or a chromosome segment and are applied to metaphase spread for the identification of translocations or aneuploidy. Repetitive probes, such as alpha satellite sequences located in the centromeric regions of human chromosomes, are used in the identification of marker chromosomes and aneuploidy. Unique sequence probes are single clones or a series of overlapping clones corresponding to a specific gene or a confined region of a chromosome that do not contain major repetitive sequences and are used for the identification of specific translocation events in cancerand for the detection of submicroscopic deletions.

#### Microarray comparative genomic hybridization<sup>7</sup>

• Recently, array-CGH (microarray comparative genomic hybridization) is considered to be useful in detecting genomic imbalance in the fetus (duplications/deletions).

#### Molecular Genetic Techniques<sup>7</sup>

Molecular genetic techniques are being used for prenatal diagnosise. The following molecular biologic techniques can be used for prenatal diagnosis of different diseases.

- Linkage analysis by microsatellite markers
- Restriction fragment length polymorphism
- Single nucleotide polymorphisms
- DNA chip
- Dynamic allele-specific hybridization

## CONCLUSION

Happiness lies, first of all, in health. Hence after getting confirmed about pregnancy it is time to take care of health and go for regular check-ups. During the 10th week of pregnancy woman has to be registered for herself and childcare, at the same time the first check-up is also conducted. It would be better if both pregnant woman and her spouse go together for these check-ups. These check-ups include knowing woman's background/history, examination and various laboratory tests. Repeated check ups are necessary to monitor maternal health, development of foetus and identify any risk causing factors like high blood pressure.

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