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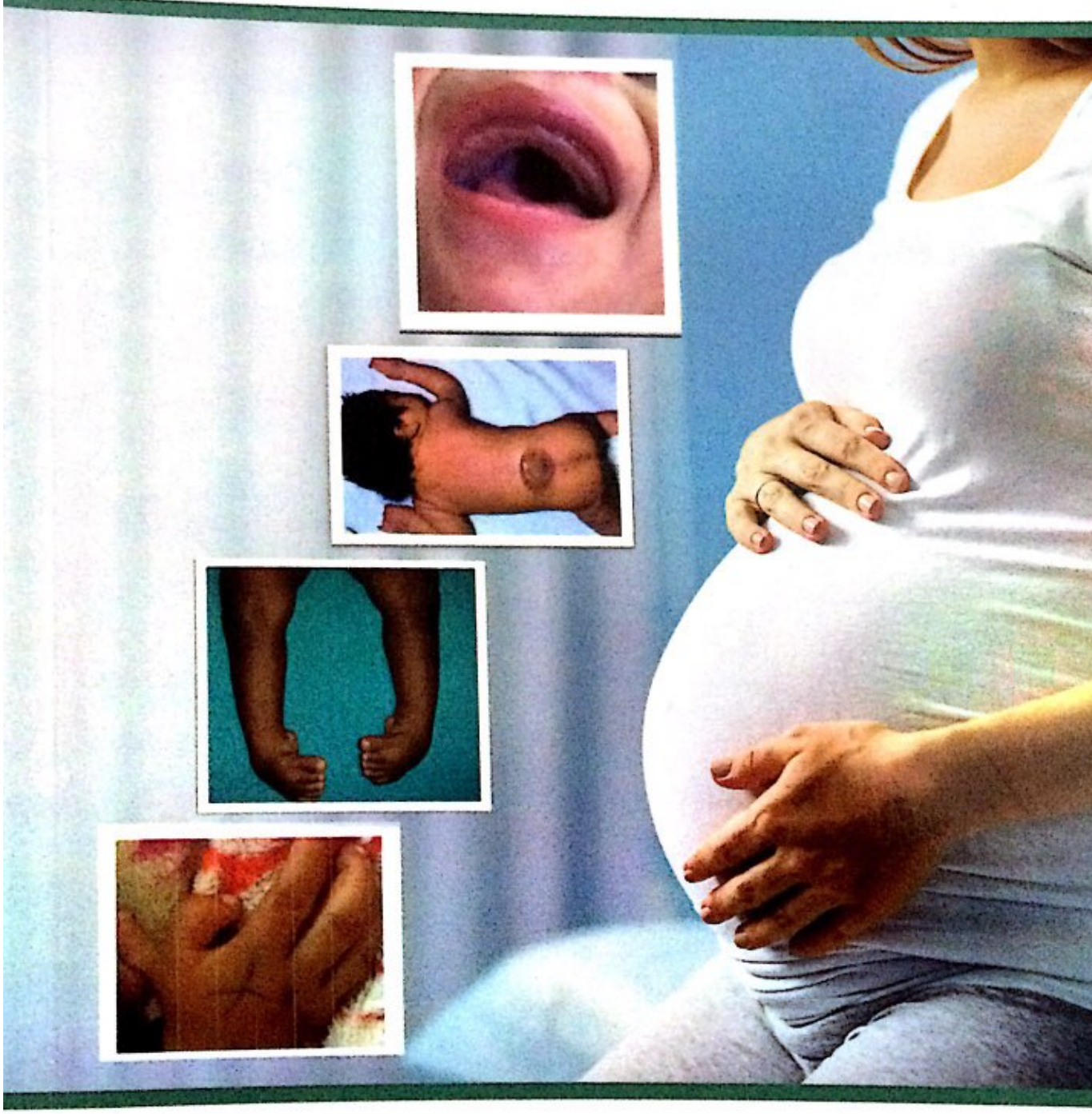
PREVENTION AND MANAGEMENT OF BIRTH DEFECTS



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Prevention and Management of Birth Defects

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Preface

Birth defects account for a significant proportion of perinatal, neonatal and child mortality. Tho children who survive may have to face varying degree morbidity and disability for the entire li India has largest numbers of infants born with birth defects in the world. This booklet will help-

a) Parents who have history of some birth defect in their previous child and want understand effect of birth defect on their future pregnancies.

b) Mothers who have a birth defect baby in their womb and want to know about vario detection and diagnostic methods as well as prognosis & management options available deal with the problem of birth defects.

C) Couples who are planning for pregnancy. Both, who have any family history of some bir defect and those who don't have any such history would be benefitted from this booklet.

This booklet is a part of Ph.D. thesis work of Dr. Alka (Research Scholar, Centre for Public Healt Panjab University) titled" Devising and validating a counseling protocol for couples reporting a OBG (Obstetrics and Gynecology) Department of PGI, Chandigarh with problems related t congenital disorders in their children". It was used as a part of an interventional package given to th couples who had a baby with birth defect.

Feedback and valuable comments of the experts in the field of gynecology, pediatrics, and Publ health are welcome for this booklet. We have tried to cover all the aspects related to birth defect Hopefully, the reader gets adequate and sufficient information on the problems faced by them.

The authors express sincere thanks to all the experts, guides and co-guides who have helped i formulation of this booklet. A special thanks to Dr. Anupriya, Assistant Professor, Department o Pediatrics, PGIMER and Dr. Bharti Sharma, Consultant, NBBD, WHO Project for their valuable comments and suggestions. We thank the parents who allowed us to use photographs of the children for the welfare of others. We also thank Century Publications for bringing out this booklet in the present shape. Support from ICMR through SRF grant to Dr Alka for doing PhD on this subject is also sincerely acknowledged

Foreword

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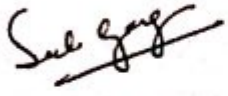
Every year around 6% of total births are born with birth defects e.g. congenital heart disease, neural tube defects and Down's syndrome followed by hemoglobinopathies. The Common risk factors associated with birth defect are genetic factors, maternal conditions and environmental exposures. Many of them are preventable and effective interventions are available like pre-conceptional folic acid supplementation, vaccination against rubella, fortifications of food with micronutrient, management of syphilis and diabetes, controlling use of toxic chemicals among others. Timely prenatal diagnosis of birth defects is essential, because in case termination is to be planned in India, this is legally allowed only up to 20 weeks of pregnancy.

Here, it is also important to understand that the nature of genetic diseases is quite different from infectious diseases. For example, typhoid or cholera occurs when people consume contaminated from or water; malaria or dengue occurs through mosquito bites, noncommunicable diseases like cancer of the tongue may occur when tabacco (khaini) is taken for many years and so on. In contrast, occurrence of birth defects or genetic disorders would also depend on whom we marry, e.g., like thalassemia, hemophilia, etc. may run in families. If we know the genetic makeup of parents it is possible ot predict how likely it is that child will have this disease. Although some tests are available now which help us in diagnosis of such diseases during pregnancy. Avoidance of consanguineous marriages is on of the method of their prevention.

This is an easy to follow booklet. It will really help those patients who due to socia-cultural barriers cannot communicate theirproblems with doctors. This booklet would help those who are planning pregnancy and have a family history of birth defects or a previous

history of multiple abortion or still birth. This would guide the parents to understand the impact of birth defects, risk factors, screening, diagnostic tests and management options available in our country. This booklet would surely help in providing pregnant women the care they need, answering the queries they have regarding congenital malformations, so that the newborn can have the best possible start in life. It would increase their current knowledge and practices related to a healthy pregnancy.

This will surely prove to be an effective empowerment tool for parents battling birth defects, congenital malformations or genetic disorders their children. I congratulate the authors for undertaking this generous task. I with them success.



(Dr. Suneela Garg)

Situations a genetic counselor may have to deal with

Parents may come to a genetic counselor in the following situations -

- H/o CMF in previous pregnancy e.g. Down syndrome
- Family history of some congenital malformation.
- H/o still birth
- Unexplained neonatal death
- H/o repeated abortions
- H/o Rh incompatibility
- Diagnosed birth defect in present pregnancy (USG shows some defect or Positive screening test results of dual/triple/quadruple test etc.)
- Diagnosed birth defect in the newborn.

Depending upon the condition a counselor may advise them about various tests available, their interpretation, risk factors associated with the condition, available treatment options available and prognosis of the continuation of the pregnancy.

Glossary of terms

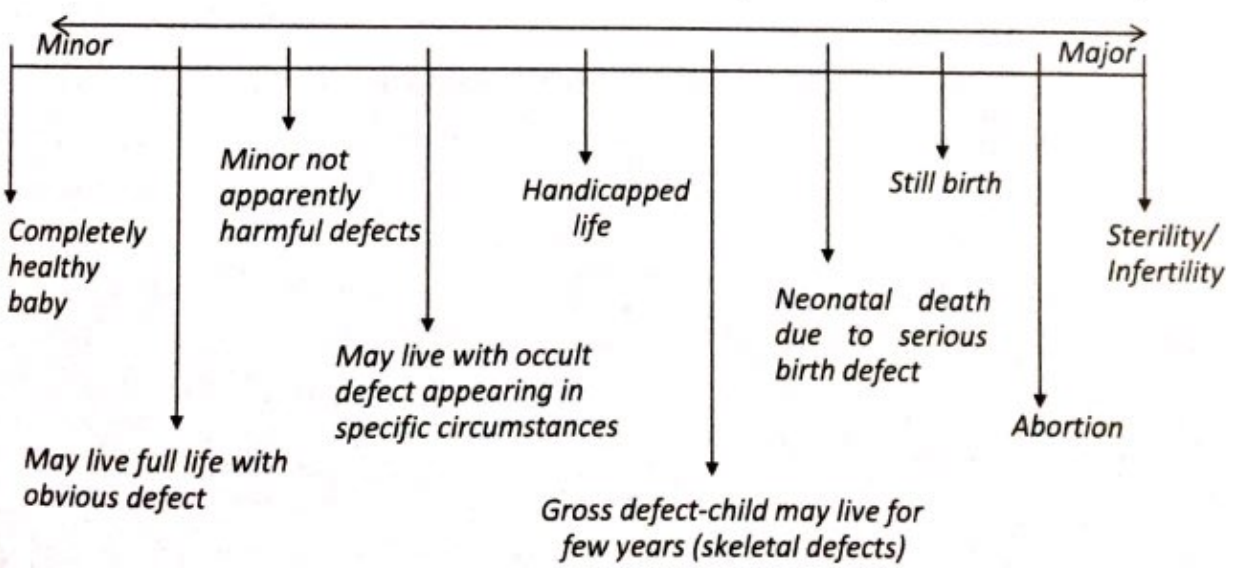
CVS	Chorionic Villus Sampling
NT	Nuchal Translucency
NTD	Neural Tube Defect
GD	Genetic Defect
CMF	Congenital Malformation
HIV	Human Immuno Deficiency Virus
NIPT	Noninvasive Prenatal Testing
VDRL	Venereal Disease Research Laboratory
STI	Sexually Transmitted Infection
TPO	Thyroid Peroxidase
CHD	Congenital Heart Disease
OPD	Outdoor Patient Department
POG	Period Of Gestation
DNA	Deoxy Ribose Nucleic Acid
SSD	Sex Selection Drug

Index

Introduction

Marriage is an important event in our society. It helps in the perpetuation of our lineage. A family is considered to be complete with the arrival of a child.Most couples have a normal healthy baby some have a baby with birth defect. After marriage, a couple may have problems like :

- Not having a child.
- Abortion may occur in approximately 15 to 20% of pregnancies.
- Some cases are diagnosed with birth defect in womb.
- In some cases, pregnancy continues up to the 9th month, but sometimes serious problems **(birth defect)** ** arises, and a dead baby may be born (still birth) due to anencephaly (no head is formed), spina bifida (gap in back bone).
- Sometimes the baby may die within hours or days, e.g., congenital heart defect (baby turns blue).
- In some cases,baby survives for a month or years e.g. cleft lip/cleft palate (split in lips or *taloo*).
- In other cases, child survives for a long time with treatment e.g. thalassemia.
- Others survive for variable periods but the quality of life is seriously compromised whereas in others it affects less, e.g., Down's syndrome (mental retardation).



Spectrum showing impact of congenital disorders

Thus, in the spectrum for the fate of any marriage with child bearing, on the one hand is the birth of a completely healthy baby & on the other hand is infertility (childlessness).

Understanding congenital disorders and related terms

Congenital disorders is a broad category that includes in itself a variety of conditions. The word congenital means 'since birth' and "disorders" means a group of diseases.

The terms congenital physical anomaly, congenital malformation, birth defects or genetic disorders are often used interchangably. However, they have different meanings.

Congenital physical anomaly: It refers to abnormality of structure of a body part" e.g. curvatures of 5th finger (clinodactyly).

Congenital malformation: It is a congenital physical anomaly that is harmful, i.e., a structural defect perceived as a problem.

Birth defect : It happens while a baby is developing in the mother's body. It is recognized at birth. It is significant enough to be considered as a problem,.e.g, cleft lip, cleft palate.

Genetic disorders: These are caused by one or more abnormalities in our genes, e.g. haemophilia A

❖ **In this book all these terms are used interchangeably.**

Types of birth defects

Birth defects may be fatal (causing death) or non-fatal (does not cause death). Anencephaly is a serious birth defect in which a baby is born without parts of the brain and skull. It is a type of neural tube defect (NTD). Syndactyly (wherein two or more digits are fused together, like HRITHIK) is nonfatal.

Birth defects can be grouped in to Major/Minor and Structural /Functional related defects.

Major defects- They can have a serious, adverse effect on the physical health, development, or functional ability of the baby, e.g., congenital cardiac defects. There are 8 externally visible major birth defects e.g. neural tube defect including microcephaly, orofacial defects (cleft lip/cleft palate), talipes equinovarus, club foot), limb reduction defects, hypospadiasis, exomphalos/omphalocele, gastrochisis, imperforate anus.

Minor defects- These include the minor variations of normal morphological features of little or no known medical, surgical, or cosmetic significance. But they can have significant psychological impact e.g. Preauricular tags, low-set ears, single transverse palmar crease etc.

Body structure related birth defects

There are two other main categories of birth defects.

These are related to a problem with body part or body structure. In short in this a specific body part is defective.

Examples - Cleft lip or cleft palate, heart defects such as hole of heart, Abnormal limbs such as a clubfoot, Neural anomalies such as spina bifida, and problems related to the growth and development of the brain and spinal cord.

				
Syndactyly	*Absent metacarpal bones*	*Right foot CTEV*	*Cleft palate*	*Spinal diastasis*

Body Function or Developmental birth defects

These are related to a problem with working of a body part or body system. These problems often lead to intellectual and developmental disability

Type	Explanation
Nervous system or brain problems	Behavioral disorders,Speech or language difficulties, Seizures and movement trouble, Down syndrome, Fragile X syndrome.
Sensory problems	Hearing loss and visual problems (blindness or deafness)
Metabolic disorders	Biotinidase deficiency, Hypothyroidism
Degenerative disorders	Muscular dystrophy , X-linked adrenoleukodystrophy

Table -1 Some examples of body function or developmental birth defects

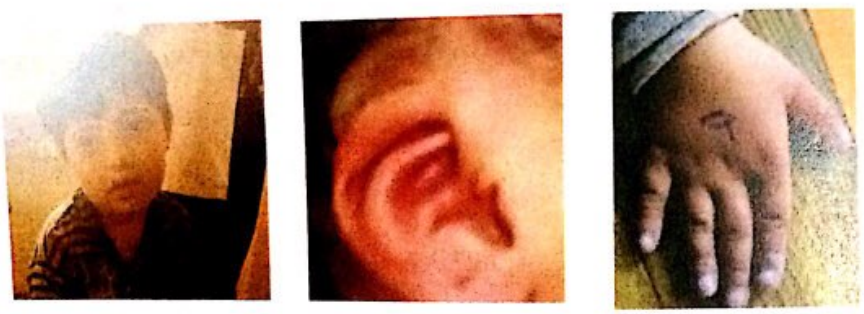


Image- Features of Down Syndrome- face / tops of ears may slightly fold over/short finger

In some cases, birth defects are caused by a combination of factors. Some birth defect affect many parts or processes in the body, leading to both structural and functiona problems.

Some examples of birth defects	What happens in this birth defect
CHD	An abnormality in the heart that develops before birth.
Down's syndrome	An extra chromosome 21 causing developmental and intellectual delays.

Some examples of birth defects	What happens in this birth defect
Cleft lip and cleft palate	Openings or splits in the roof of the mouth and lip.
Spina bifida	A birth defect in which a developing baby's spinal cord fails to develop properly.
Club foot	A birth defect in which the foot is twisted out of shape or position.
Phenylketonuria	A birth defect that causes an amino acid called phenylalanine to build up in the body.
Cystic fibrosis	An inherited life-threatening disorder that damages the lungs and digestive system.
Huntington's disease	A condition in which nerve cells in the brain break down over time.
Duchenne muscular dystrophy	A disorder of progressive muscular weakness manifests in children, typically in boys.
Sickle cell anemia	A group of disorders that alters shape of red blood cells.
Hemophilia	A disorder in which blood doesn't clot normally and bleeding in joints occurs (especially in boys)
Thalassemia	A blood disorder involving lower than normal amounts of an oxygen carrying protein, requiring blood transfusion.

Current Scenario and Burden of Birth Defects

The birth prevalence of congenital anomalies in the developing world is underestimated. This is due to deficiencies in diagnostic capabilities and lack of reliability of medical records and health statistics. As a result, recorded diagnoses in vital statistics focus on overt acute illnesses, rather than on pre-existing congenital conditions that increase vulnerability to infections and malnutrition (WHO, 1985). Every year 15 lakhs children (6% of total births) are born with birth defect in India. Major birth defects are diagnosed in nearly 6% of the infants. Congenital anomalies affect 1 in 33 infants.There are 3.2 million birth defect related disabilities.Birth defects associated deaths; predominantly occur in first 24 hours; up to 7 days of life.Every year approximately 2.7 lakh newborn die before 28 days of life due to birth defects.70% of the BD's are treatable and manageable if identified at birth. Most common serious congenital disorders are heart defect,neural tube defect and Down's syndrome.

Amitabh Bachchan playing the role of a child who is suffering from Progeria, a genetic disorder which leads to quick acceleration of ageing process in children

Hrithik Roshan has two thumbs on his right hand - and it concerns a rather complex combination of abnormalities: 'polydactyly' (= extra digits) + 'syndactyly' (= fused digits).

Varun dhawan as conjoined twins after surgery in Movie judwaa 2

Late Bollywood actress Madhubala suffered from congenital heart disease. She had a Ventricular Septal Defect (VSD), a disorder referred to as a "hole in the heart." At the time of her birth, there was no effective treatment available for VSD. If this would have been available she would have lived to celebrate her 80th birthday today with us. Bollywood star Hrithik Roshan has an unusual thumb: he has two thumbs on his right hand - and it concerns a rather complex combination of abnormalities: 'polydactyly' (= extra digits) + 'syndactyly' (= fused digits).In her recent post on facebook, actress Celina Jaitley on September 10, had talked about her twins and their birth. She revealed the bitter news of having to lose one child due to a congenital heart defect.

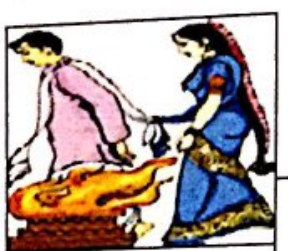
Risk Factors

Risk of having a child with birth defect increases under any of the following conditions:

Inadequate Periconceptional intake of folic acid

Women giving birth after 35 years of age

Use of teratogenic medications



Consanguineous marriage

Risk Factors

Overweight

Lack of vaccination against Rubella

Iodine deficiency in mothers diet

Alcohol intake and smoking

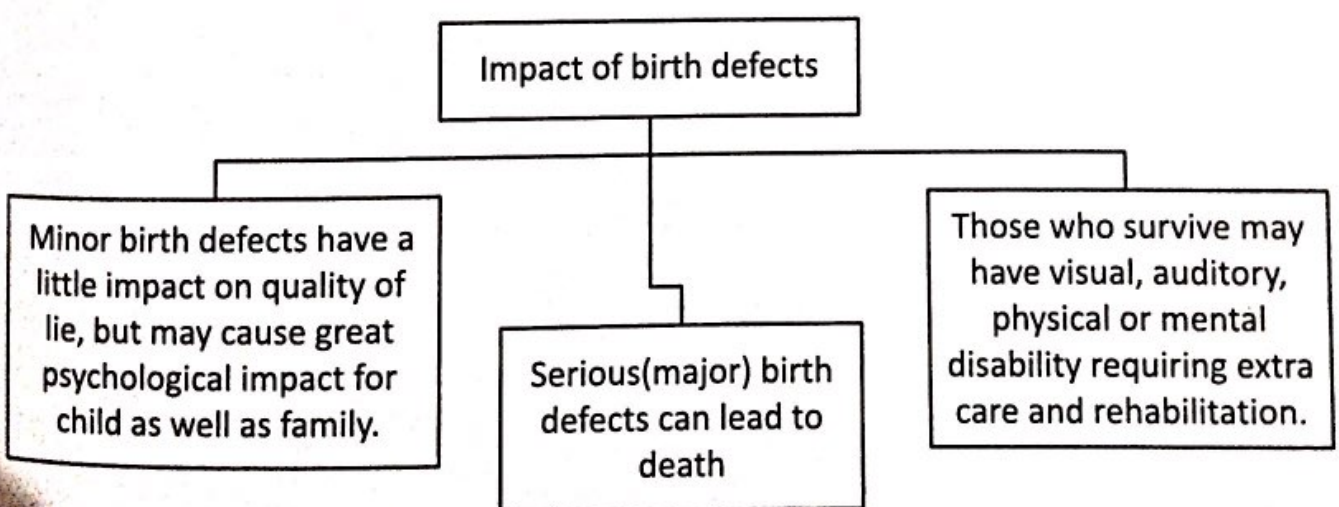
Impact on Quality of Life

Having a child with a birth defect impacts various spheres of life of children as well as family.Birth defects may lead to mental,social,medical, family problems due to long term suffering for the patient.

Parents of children who have birth defects face unique challenges and they desire to make life better for their kids. They need psychological support.

They also want to understand the way that can help them to prevent birth defects in their future children. Some of the challenges parents face involve communication with healthcare professionals.

There are also quality of life issues. The treatment of some genetic disorders or birth defect scan be quite expensive and require government support or other sources of funding in selected cases.



Why do birth defects occur?

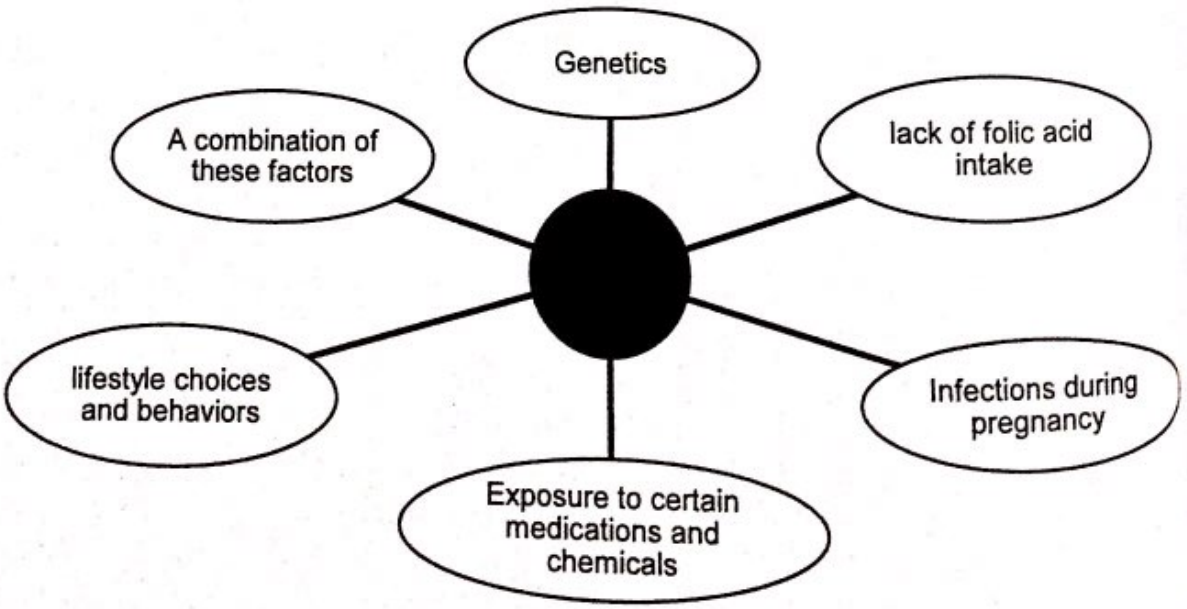
"HE is the one who shapes you in the womb as He Wills -Quran"

Every culture views life experiences differently; therefore, each ethnic or racial group ha different beliefs and theories of causation of illness. Many of the nonmedical causes of birt defects are attributed to maternal impressions, something the pregnant woman thinks o sees e.g. a child born with microcephaly or anencephaly is the result of his/her mothe looking at a monkey during the pregnancy. Some believe that a cleft lip is caused by lookin at or eating a rabbit, hence the name"hare-lip".

Some cultural groups believe persons with supernatural powers can also cause birt defects. In some cultures, individuals may feel that a family is "given" a child with a disorde as a punishment by God for parental sin. The "evil eye, is a force that gets much recognitio in many cultures for being the determinant of bad fortune, including birth defects.

There are many other examples of superstitions such as :the effect of the moon, specially lunar eclipse causes cleft lip or palate, spina bifida can result from eating potato eyes durin pregnancy, eating chili peppers in pregnancy may cause blindness in the fetus .

As per medical sciences the birth defects can be due to many factors, However, the exa causes of certain birth defects are often unknown.



Causative Factors

Birth defects may be caused by genetics or by environmental factors or a combination of both. In certain disease genetic element is so overwhelming that it expresses itself predictably without any need of environmental challenge.

Genetic (Hereditary)

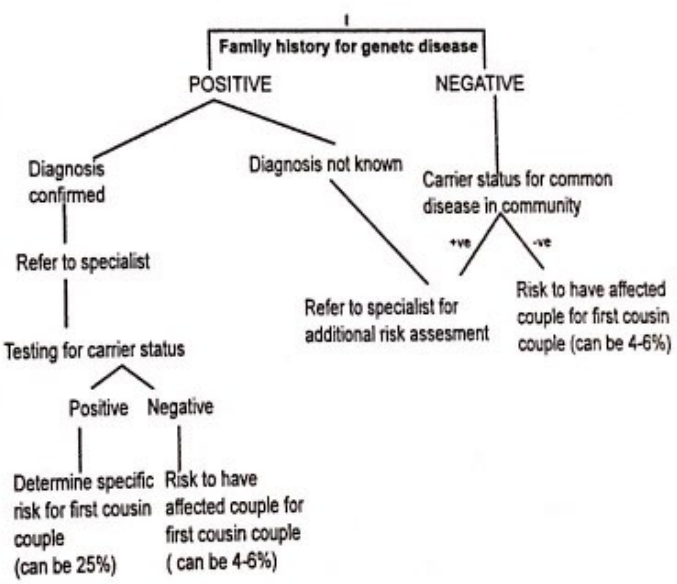
The mother or father may pass on genetic abnormalities to a baby.These defects are caused by mutations or consanguinity and are unpreventable. There are three categories of genetic disorders: chromosomal disorders, multi-factorial inheritance and single-gene disorders.

Mutation

It means abnormality in the gene, leading to disease. It also includes the new mutations in one of the germ cells that gave rise to the fetus.

Consanguinity

This refers marriage among close blood relatives such as first cousins and uncle-niece marriages. Endogamy (practice of marrying within relatives/cousins or a specific social group, class or ethnic group) should be avoided and exogamy should be preferred. Such marriages increase the probability of mating between two individual heterozygotes for the same recessive gene. The offspring of consanguineous parents are at a risk of many diseases like cancer, mental disorders, beta thalassemia, hypertension, hearing deficit, and leukemia, diabetes mellitus, and epilepsy, congenital heart diseases. The practice of preferential cousin marriages has been recorded as a characteristic feature of Muslims; in Some Hindu groups too and among the Gujjars also.



Environmental

Advanced maternal age

Risk of having a baby born with congenital malformations increases with increase in age of mother. e.g. Down's syndrome (Age affects quality of sperm and ovum).

Alcohol use

Consumption of alcohol by the mother during pregnancy can cause many birth defects e.g heart disease, brain damage, kidney abnormality, behavioral and cognitive disorders like hyperactivity including difficulties with learning and memory.

Paternal smoking

Smoking prior to conception can lead to some mutation in DNA of the parent. Hence there may be an increased risk of passing congenital abnormality in the offspring.

Maternal obesity

Obesity has been found to be linked with an increased risk of certain types of birth defects This may include neural tube defects, shoulder dystocia, Macrosomia.

Toxic substances

Toxic substances that can cause congenital disorders are called "teratogens". These may include usage of certain drugs or exposure to many environmental toxins during pregnancy. Examples-intake of valpoic acid by pregnant lady in the first trimester may lead to heart defects, cleft lip, or a neural tube defect (an opening in the baby's spine or brain). Other examples may include use of Phenytoin (in epileptic women) and thalidomide (used to treat morning sickness during pregnancy). Even a single dose caused severe birth defects in first trimester, including amelia (absence of limbs), phocomelia (short limbs), incomplete or absent bone growth, ear and eye abnormalities, congenital heart defects.

Baby born to a mother who had taken thalidomide in pregnancy for multiple myeloma

Infections

Infection caused by some virus ,bacteria, or, parasites get transmitted directly from the mother to the embryo, fetus or baby during pregnancy or during childbirth. Fetal exposure to rubella during 5-10 weeks of development (the sixth week particularly) can cause cataracts in the eyes. It can also cause abnormalities of the internal ear, heart, and sometimes the teeth also. Other infectious agents may be syphilis, herpes simplex virus, toxoplasmosis, etc.

Radiation

Excessive exposure to radiations can be harmful for both the mother and her unborn baby. The mutations caused in the DNA by radiations can lead to birth defects e.g. reduction in height, mental retardation, small head size and retarded brain development. One of the well-known harmful environmental radiation hazards is Chernobyl disaster(reactor explosion) that occurred in Pripyat, Ukrainian SSR, Soviet Union on April 26, 1986. According to the Chernobyl Children International (CCI) lives of millions have been affected and many have been found to suffer from congenital heart defects and other disabilities too.

Lack of nutrients

Lack of folic acid in the diet of a mother can cause neural tube defect (NTD) and Spina bifida. 5 microgram of folic acid, before the pregnancy and till 12 weeks of pregnancy can help in prevention of some birth defect.

Unknown or multifactorial

Approximately 65% of the birth defects have no known cause. This shows their random occurrence regardless of maternal living conditions.

Some important chemicals that can lead to birth defects

Chemical	Source	Effect
Lead	Lead-based paint or occupational exposure	Premature delivery, abortion/ miscarriage, Low birth weight Neurological damage, Developmental delays
Methyl Mercury	Fish and seafood	Effects fetal nervous system development
Pesticides	Unwashed vegetables and fruits, contaminated water	Increases the risk of birth defects
Toluene	Shoemaking, painting, printing, Varnishes, gasoline, glues.	Increased risk of abortion and low birth weight.
Carbon monoxide	Gas fumes, automobile and industrial exhaust, cigarette smoke	Low birth weight and premature delivery.
Organic solvents	Alcohols, paint and paint thinners, nail polish removers, paints, varnish removers	Spina bifida, heart defects, clubfoot, and deafness
Glycol ethers	Photographic applications, silkscreen printing, and dyes.	Increased risk of miscarriage
Chlorine	Chlorinated drinking water	Increased risk of miscarriage and poor fetal growth.

Use of Desi- Dawai (Herbal /Local Medicines) During Pregnancy

Use of desi –dwai during pregnancy to ensure birth of a baby boy is an important cause of birth defect. Consumption of indigenous medicines used for sex selection, also known as Sex Selection Drugs (SSD) during the first trimester of pregnancy is prevalent in certain northern parts of the country.

It is important to note that SSDs are consumed during the most critical period of development of the embryo, i.e., first trimester of pregnancy to have a male baby. Consumption of such drugs is dangerous and detrimental to the growth and development of the embryo. These contain phytoestrogens and testosterone. Shivlingi & majuphal are the herbs used for such purpose. These are often sold in tablet or powder forms. Often mothers in law or other relatives force the pregnant women to take such medicines to get a son.

Use of such medicines may lead to an unfortunate situation, where the baby is born which looks like male (external genitals male, but actually it is a hermaphrodite i.e. a hijra. So there is no benefit (rather harm).

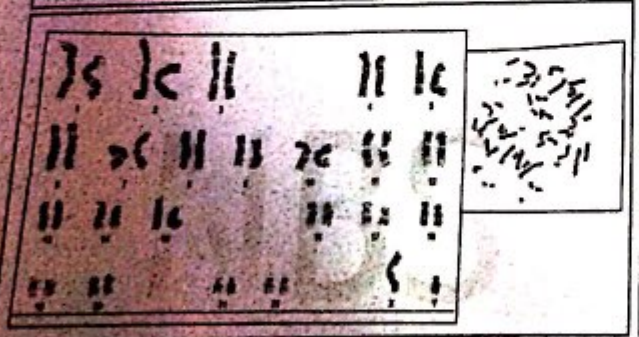
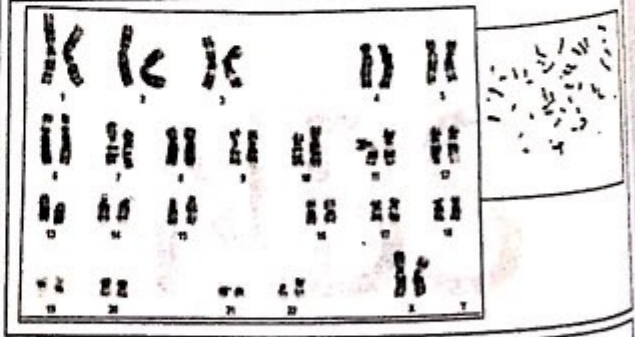
Research from PGIMER, Chandigarh by Dr. Sutapa, Dr. Chinmayea and Dr. Amarjeet Singh (School of Public Health) revealed that more than 90% of the women were aware of the availability of SSDs. Fifty (45.5%) of them reported to have used these drugs. Use rate of SSDs was significantly more in women who gave birth to a child with birth defect as compared to those who didn't.

What to do if Parents have a baby with congenital disorder

Every child born with birth defect needs immediate neonatal screening e.g. In baby born with G6PD deficiency, neonatal jaundice & kernicterus may appear, affected infants are monitored for several weeks for bilirubin level. Early management may help a child to lead a better life. e.g. a baby born deaf if not diagnosed early will have dealyed speech, as child wont be able to hear to learn to speak. They are following two situtions that arise when a couple already has a child with birth defect.

What to do in preconceptional phase? What to do in antenatal phase ?

Preconceptional phase-Before planning pregnancy parents should go for preconceptional counseling. During this the risk factors will be assesed and the tests required to confirm the diagnosis of birth defect in child will be done through neonatal screening. They may be referred to geneticist in pediatrics for diagnosis. e.g If a couple already has a child that represents features of down syndrome, karyotyping is must to confirm the diagnosis before planning for next pregnancy. Although neonatal screening generally plays a smaller role than antenatal screening, still it provides an important opportunity to identify birth defects early in the neonatal period. This way parents may be able to seek therapy or surgery for

Karyotyping Report of Father	Karyotyping Report of Mother
Karyotyping on Blood Samples	Karyotyping on Blood Samples
No. of Cells Counted : 20 Estimated Band Resolution : 450 Number of cells Karyotyped : 05 Banding Method : GTG	No. of Cells Counted : 20 Estimated Band Resolution : 450 Number of cells Karyotyped : 05 Banding Method : GTG
	
Reason For Referral : Previous Baby with multiple Malfunctions. Karyotype : 46,XY Results : Chromosomal analysis of PHA stimulated cultured lymphocytes revealed an appartently normal Male Karyotype with no structural and numerical abnormalities. Interpretation : Normal Male Karyotype	Reason For Referral : Previous Baby with multiple Malfunctions. Karyotype : 46,XX Results : Chromosomal analysis of PHA stimulated cultured lymphocytes revealed an appartently normal Female Karyotype with no structural and numerical abnormalities. Interpretation : Normal Female Karyotype

child early in life. Early and appropriate treatment can prevent or reduce some lethal or disabling sequelae of birth defects. For neonates born in hospitals, screening should occur before they leave. Even when little can be done to help the infant, accurate diagnosis of birth defects can alert parents to the risks they may face in future pregnancies.

Antenatal phase-In case a couple has not gone for genetic counseling in preconceptional phase then in antenatal stage both neonatal screening and antenatal care/genetic counseling to rule out any CMF is must.

e.g. if couple already has a a thalasemic child then at 11-13 week current pregnancy they are advisedto go for chorionic villi sampling (CVS) to confirm the same in fetus. If needed thay are also referred to a genetic counselor. Depending upon the gestatonal age they are advised to decide regarding fate of fetus.(MTP/ Continautionof pregnancy).

Genetic counselor will help them to understand the

- Available screening tests, diagnostic methods, and options for preventing or treating birth defects.
- Limitations of screening tests are explained, along with the goal of identifying high-risk pregnancies.
- Results of the screening process and answers their questions. Those who had a positive screening test will be provided information on the diagnosis, etiology, prognosis, and consequences.
- Impact of that birth defect on baby's health.
- Educate them about reproductive choices, there by allowing them to make free and informed decisions.
- Risk of having a baby with birth defect in the next pregnancy.

Investigations Available for Detection and Diagnosis of Birth Defects

There are many birth defects that can be detected during pregnancy with early screening. Timely detection can give a chance for medical termination of preganancy (before 20 weeks of pregnnacy). But if it is detected after 20 weeks MTP cannot be done as per PCPNDT act in India. At this time a multidisciplinary approach should be followed. Prognostication should be done. Parents must be completely informed about the diagnosis. They must be given psychological support and should be motivated for institutional delivery e.g. in case of diagnosed CHD if a baby is delivered at a tertiary care hospital ,immediately baby may be given cardiology care because he may need a pacemaker.

If a antenatal women who is at risk for birth defect somehow misses these detection tests baby may be born with a birth defect. However these tests does not give 100% surity of detection of all the birth defects some birth defects can only be diagnosed after birth of the baby.

Screening test

With early Prenatal Screening & detection it is possible to diagnose a birth defect as early as possible during pregnanacy. Two types of tests are available. Screening test and diagnostic tests.

- **For whom are prenatal screening tests recommended?**

  These are recommended for all expectant mothers.

- **How are screening tests performed for genetic disorders?**

  Both, Invasive and noninvasive (ultrasonography) tests are available for the screening. These clearly distinguish between individuals who are at risk for the condition and those who are not.

- **What happens if screening tests show that there is a problem?**

  Positive test results usually indicate a higher chance of having a particular genetic disorder. This leads to consideration of further diagnostic tests.

These provide individual risk assessment tests, examinations or other procedures, e.g.,

Down's syndrome or a neural tube defect. A positive report only hints at the possibility of occurrence of a birth defect. If needed, a diagnostic test is done to confirm the presence of a genetic disease.

Diagnostic tests

Genetic screening of populations identifies clinically normal individuals who have genotypes associated with a birth defect or who are at high risk of producing offspring with a birth defect. It aims to identify as many affected individuals as possible, but screening alone does not detect all individuals at high risk. Diagnosis usually follows a positive screening test.

Diagnostic tests are usually recommended when a screening test has shown an increased risk of a birth defect. These are the tests that definitively identify a defect if present,e.g, Chromosomal abnormality (aneuploidy), genetic mutation (Sickle cell), etc.

- **For whom are prenatal diagnostic tests recommended?**

When a screening test is positive and also when couples already have a child with genetic disorder (e.g. previous baby with down syndrome) that increase the risk of having a baby with certain birth defects.

- **How are diagnostic tests for genetic disorders performed?**

Diagnostic tests are done on the cells obtained from the fetus (amniocentesis or chorionic villus sampling (CVS)). These cells are analyzed using different techniques, e.g, Karyotyping

- **What happens if diagnostic tests show that there is a problem?**

If a diagnostic test shows a positive result, a multidisciplinary approach helps in making choices and considering the available option.

List of various tests

Screening test	Diagnostic tests
• Thalassemia Screening	• Chorionic villus sampling
• Thyroid Screening	• Amniocentesis
• Nuchal Translucency (NT& NB Scan) +USG	
• Dual Marker+ UGG	

- Triple Screen/Quadruple Screening,
- NIPT

Thyroid Screening

Thyroid problem can harm the baby as well as the mother in pregnancy. During pregnancy two pregnancy related hormones HCG and oestrogen increase thyroid hormone levels in blood. It is critical for normal development of brain and nervous system of the fetus. During first trimester, supply of thyroid hormone to fetus is through placenta of mother. Baby's thyroid begins to function at its own after 12 weeks of gestation.Hypothyroidism results in misscarriage, preterm births (in 7th or 8th month), intrauterine growth restriction, and fetal death inside uterus, problem with breathing or heart beat and increased perinatal mortality.

Pregnancy specific and trimester specific reference levels for TSH are as : Ist trimester - 0.1-2.5mIU/l; IInd trimester - 0.2-3mIU/l; IIIrd trimester - 0.3-3mIU/l. venous blood samples should be taken with other antenatal care (ANC) investigations in a single sitting

Thalassemia Screening

All the antenatal mothers are advised for thalassemia screening through Hb electrophoresis between 10-12 weeks of pregnancy. If the result of the test is positive, then the partner is advised to go for thalassemia screening. If the result of test is negative, then nothing is to be done. But if the result comes out to be positive in both the partners, a diagnostic test (Chorionic villus sampling) is done to confirm thalassemia in fetus.

If the baby is affected by alpha thalassemia major, couple is advised to go for MTP. The time for undergoing the test is crucial because if the fetus turns out to be thalassaemic, MTP has to be done within 20 weeks of pregnancy.

First trimester screening for birth defects

First trimester screening includes a test of pregnant women's blood and an ultrasound examination. Both tests are usually performed together at 11-13^{+6} weeks of pregnancy.

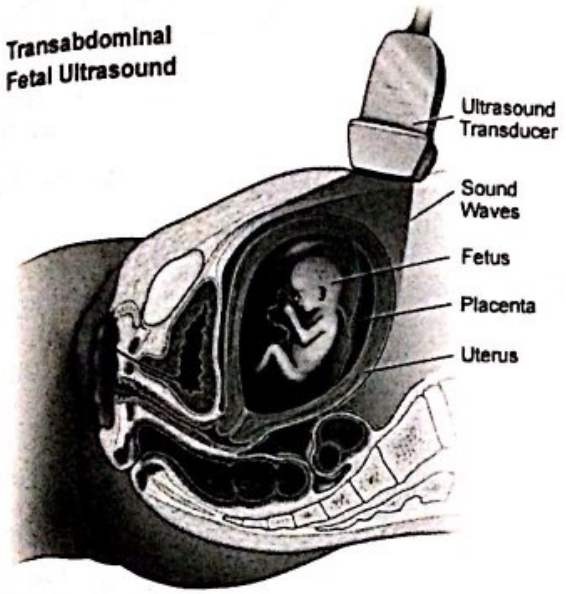
Nuchal translucency/ Nuchal bone (NT/NB) scan

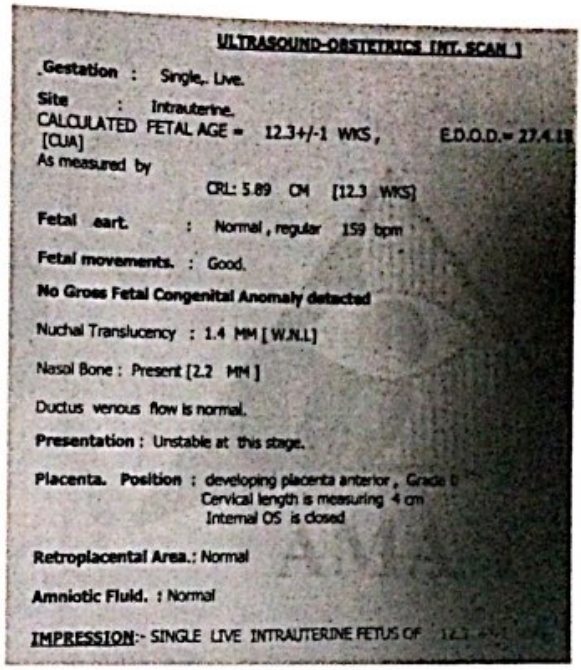
This test involves a ultrasound scan of the collection of fluid under skin at the back of baby's neck. A Value of 2.5-3.5 indicates a normal result,whereas a value greater than 3.5 indicates some problem (Down's syndrome, trisomy 18, congenital heart defects, other genetic

syndromes). Results can be obtained the same day. The measured value of NT on USG scan is assessed in relation to the period of pregnancy.

Advantages- Safe, Non-invasive.

Disadvantages-It detects only 80% of Down's syndrome cases.





ULTRASOUND-OBSTETRICS [INT. SCAN]

Gestation : Single, Live.

Site : Intrauterine.

CALCULATED FETAL AGE = 12.3+/-1 WKS, E.D.O.D.= 27.4.19
[CUA]

As measured by

CRL: 5.89 CM [12.3 WKS]

Fetal eart. : Normal, regular 159 bpm

Fetal movements. : Good.

No Gross Fetal Congenital Anomaly detected

Nuchal Translucency : 1.4 MM [W.N.L]

Nasal Bone : Present [2.2 MM]

Ductus venous flow is normal.

Presentation : Unstable at this stage.

Placenta. Position : developing placenta anterior, Grade 0
Cervical length is measuring 4 cm
Internal OS is closed

Retroplacental Area.: Normal

Amniotic Fluid. : Normal

IMPRESSION:- SINGLE LIVE INTRAUTERINE FETUS OF

Dual Marker (for 2 factors)

This test is performed on maternal blood sample. It detects free HCG and PAPP-A level in the blood. In first trimester, it identifies pregnancies that have a high risk of having babies with chromosomal abnormalities e.g. Trisomy 21 (Down's syndrome), Trisomy 13 (Patau syndrome),Trisomy 18 (Edwards syndrome). It is not a diagnostic test, hence it only identifies the risk of the baby being affected.

2nd trimester screening for birth defects

2nd trimester scrrening includes blood test (triple test, quadruple test), ultrasonography (level 2 USG) and if required ECHO.

Triple test (for 3 factors)

This is performed between 15 to 20 weeks of pregnancy. In this test blood sample is taken

from mother, it measures level of AFP, HCG, Estriol.It can detect NTD, spina bifida, trisomy 21(Down syndrome), trisomy 18 (Edward's syndrome) or other types of chromosomal abnormalities.

AFP: a protein that is produced by the fetus.
HCG: a hormone produced within the placenta.
Estriol: an estrogen produced by both the fetus and the placenta.
Triple test has a 70% sensitivity and 5% false-positive rate.

The quadruple test (for 4 factors)

This test is performed between 16 to 18 weeks of pregnancy. This test is similar to triple test. Only difference is that it tests inhibin-A(a protein produced by the placenta and ovaries) in additional to other factors of triple test. This test can detect trisomy 21 and trisomy 18 and neural tube defects. Report of the test usually comes in 2-3 days.It can predict about 85% of open neural tube defects, problems with the development of the baby's brain and spinal cord. It predicts about 80% of fetuses with Down syndrome in women over age 35 and about 75% in younger women.

QUADUPLE TEST

Maternal screen (Quaduple test) 2 nd trimester test (14 to 22.6 weeks) HCG-beta, AFP, uE3,Inhibin A

INVESTIGATION	OBSERVED VALUE	UNIT
AFP-Alpha Feto Protein (CMIA)	55.4	IU/mL
Beta HCG (Total) (CLIA)	37510	mIU/mL
E3, unconjugated Estriol (CLIA)	0.8	ng/mL
Inhibin A (CLIA)	371.2	IU/mL

Risk factor calculated by Prisca 5

Disorder	Screen positive Cutt off (ACOG 2007)	Romarks
Trisomy-21	1:250 for all age groups AFP MoM < or=0.74, HCG MoM> or= 2.06 UE3 MoM < or 0.75, Inhibib A MoM> or=1.77	Confirmatory tests needed under doctor's advise
Trisomy-18	1:100 for all age groups AFP MoM< or=0.65, HCG MoM< or=0.36 UE3 MoM < or=0.4	Level-III ultrasound needed for confirmation
Open Neural Tube Defect	AFP MoM above 2.5	Scan of Rachis recommended

Level 2 Scan

- This is a non invasive procedure (without inserting any instrument inside the

body). This is performed at around 18-22 weeks of pregnancy. It detects any gross CMF(major physical defects in the brain and spine, facial features, abdoen, heart and limbs). A TIFFA scan (Targeted Imaging for Fetal Anomalies) is also popularly known as fetal anomaly scan. It involves a detailed scanning and examination of the fetus for any abnormalities. It can be a 3-dimensional scan or 4 –dimension.

Advantage-USGs are noninvasive and have minimal risk for any complication to the fetus or mother.

Disadvantage - Unnecessary USG can put unnecessary exposure to radiation.

Some of the Problem detectable by USG	
Spina bifida	Open spinal cord
Anencephaly	Absence of the top of the head
Hydrocephalus	Excess fluid within the brain
Major congenital heart problems	Some defect in heart
Diaphragmatic hernia	A defect in the muscle which separates the chest and abdomen
Exomphalos/gastroschisis	Defects of the abdominal wall
Major kidney problems	Missing or abnormal kidneys
Major limb abnormalities	Missing or abnormal limbs
Cerebral palsy	Spasticity
Down syndrome	May be associated with heart and bowel problems

Face
Fetal face seen in profile view
Both orbits, nose and mouth appeared normal
IOD 1.1 cm
OOD 3.1 cm

Thorax
No intra thoracic mass seen.
Heart appears in the mid position.
Four chamber view normal. Echogenic focus seen in LV.
This is the basic examination of heart according to the ISUOG guidelines.

Abdomen
Stomach visualized. The umbilical vein convexity is seen to the right. Gall bladder seen between portal vein & the stomach. Normal bowel pattern appropriate for the gestation seen. No evidence of wall intact.
Hyperechoic focus measuring 6-7 mm seen below the left hemidiaphragm likely in the spleen.

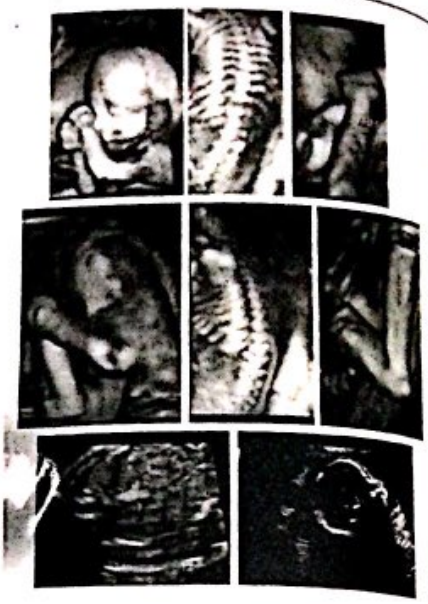
KUB
Kidneys & bladder appeared normal.

Extremities
All fetal long bones visualized and appear normal for the period of gestation.
Both feet appeared normal
There is an extra digit seen along the lateral aspect of both the hands.

Impression

Single live foetus of 20-21 weeks of gestation showing intra abdominal calcific focus, polydactyly both hands and persisted right umbilical vein.
Presently no other gross CMF seen.
Uterus showing intramural fibroid (2.9 x 3.0 cm anterior wall).
Uterine artery showing normal flow.
To correlate clinically/Suggested Fetal echo. & maternal TORCH

ULTRASONOGRAPHY



3D /4D SCAN

- **Fetal echocardiography** is a test similar to an ultrasound. It may be advised in conditions where an Unborn child is at risk for a heart abnormality or other disorder or Couple already have a child with a heart condition, Mother has b een exposed to medications that can cause heart defects(e.g.epilepsy treatment drugs),Medical conditions(including rubella, type 1 diabetes, lupus, or phenylketonuriaetc).
- **Non invasive prenatal testing (NIPT) -** In addition to above tests, NIPT (if required) can be performed in first trimester or second trimester.

Non invasive prenatal testing (NIPT)

This is done at 10-22 weeks of pregnancy, Blood sample of mother is sent to laboratory where it tests small amount of DNA of baby that is naturally found in blood of mother. It is performed for Down syndrome, trisomy 18, and hemophilia. This is a screening test. If result of the test is positive, amniocentesis (diagnostic test) is done to confirm the results.

Advantage-Detection rate for Down syndrome is better than other test

Disadvantage: It does not detect many chromosomal abnormalities.`

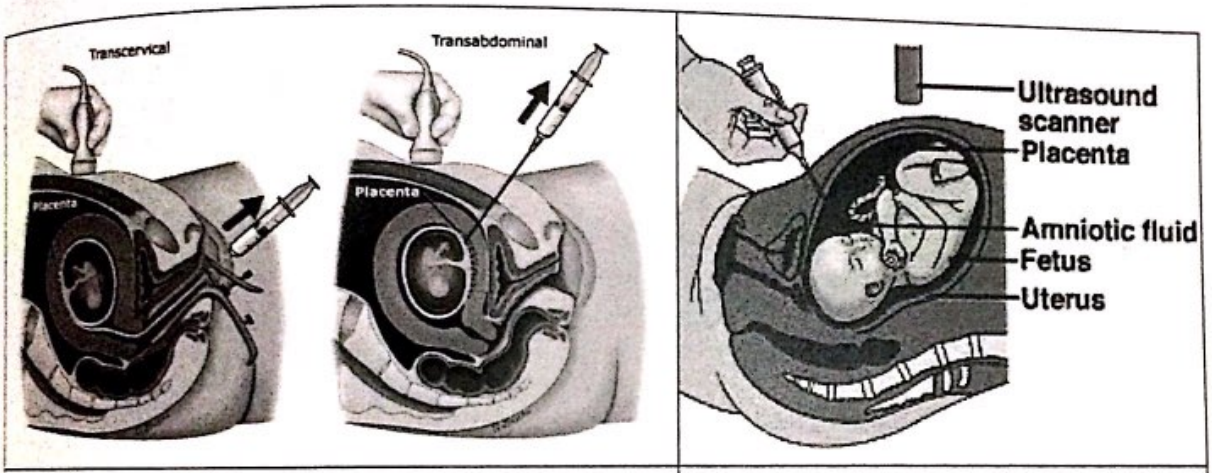
Diagnostic tests

Chorionic villus sampling(CVS)

It is done between 10^{th}-13^{th} weeks of pregnancy. In this test, with the help of a needle sample is taken from chorionic villi (placenta tissue) of the growing fetus. Then the sample is sent to laboratory for the test e.g. karyotyping or enzyme analysis for Down syndrome. Approximate timing for result is 1-2 weeks for karyotyping and 1-2 days for FISH Test.

Advantages– helps in early diagnosis and timely intervention.

Disadvantages –Procedure related loss rate is < 1%.



Chorionic villus sampling	Amniocentesis

Amniocentesis

It is done between 15-20 weeks of pregnancy. In this test sample is taken from amniotic fluid present around the growing fetus. Ultrasound is used to help doctors to show location of baby and placenta so that needle could be inserted safely to the right place. Then the sample is sent to laboratory for the test, e.g. karyotyping for trisomy 18. Approximate timing for result is 2 weeks for karyotyping and 1-2 days for FISH Test.

- **Advantages**— helps in early diagnosis and timely intervention.it is relatively safer.
- **Disadvantages** –procedure-related loss rate is approximately 0.11%. Relatively late diagnosis.

Indications of diagnostic tests- Amniocentesis or CVS is typically offered for pregnancies at increased risk for chromosomal abnormalities risks.

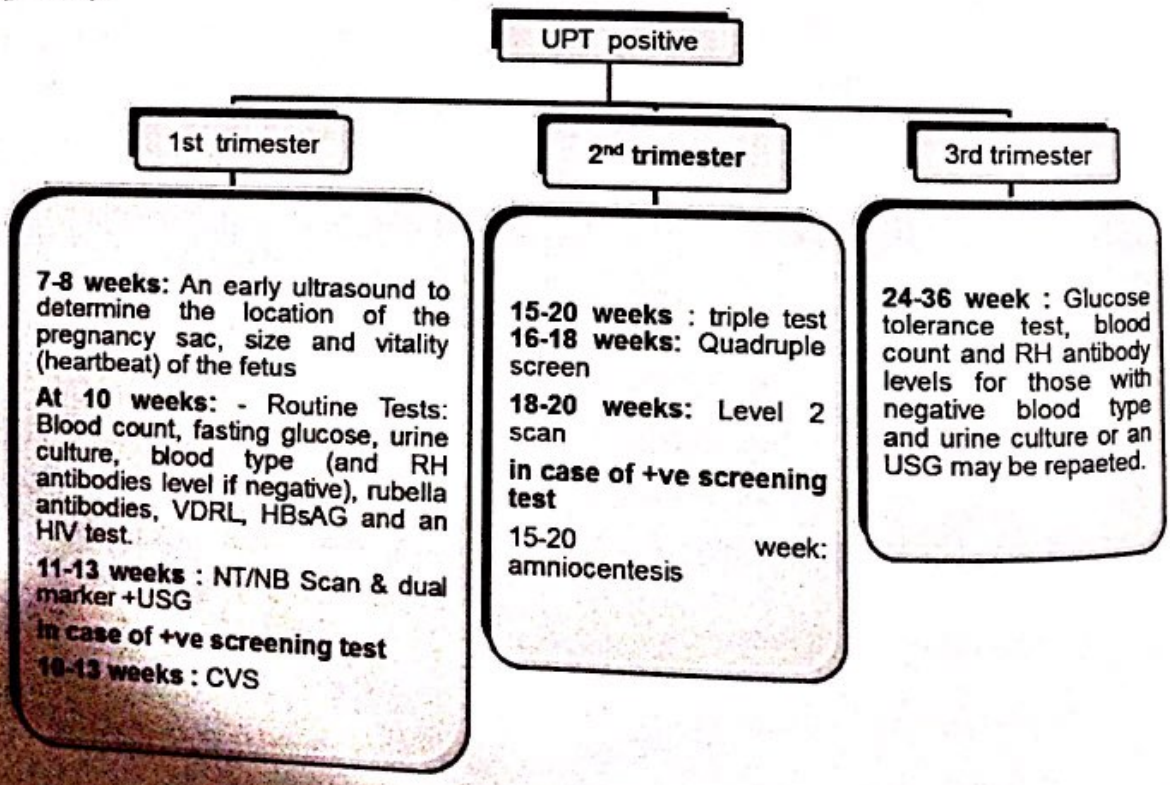
- Couple already having a baby with down syndrome.
- Any genetic disorder history e.g. both the parents are carrier of thalasemia then CVS is a must for fetus.

Routine Investigations During Pregnancy

Always try to have a planned pregnancy, so that early screening could be done and folic acid can be done timely. After concieving there are specific routine tests must be followed by a pregnant women.

Preconceptional–Before planning preganncy every women should be screened for sexually transmitted infections (HIV/VDRL), Rubella and any other medical conditions (diabetes/thyroid). Beside this complete haemogram / blood group (Rh type)/ should be done. if a women it at risk for having a birth defect baby she must go for genetic screening e.g. thalassemia screening, karyotyping(both the partners).

Antenatal-After missed period, a urine or blood test can be performed to confirm pregnancy.



Specific tests done in women who are at risk of giving birth to babies with birth defects

Test	Approximate cost		Test is done for ??
	In PGI(Rs/-)	Outsisde(Rs/-)	
Screening test			
Hemoglobin(Hb)	50 (appx.)	150 (appx.)	The hemoglobin test is used to check amount of hemoglobin in blood.
TSH	100(appx.)	280(appx.)	It is used to detect thyroid disorders
Hb Electrophoresis	350(appx.)	1050(appx.)	This can detect HbS(sickle-cell disease),blood disorders such as beta thalassaemia and hemoglobin C
VDRL(Syphilis)	50(appx.)	170(appx.)	Assesses whether or not the patient has, a sexually transmitted infection (STI) e.g.Syphilis
Dual Screen	Not available	2400(appx.)	Dual marker(Blood test for HCG and PAPP-A)+ NT For Down's syndrome
Triple screen	Not available	AFP-900(appx.) HCG-775(appx.) ESTRIOL - 750(appx.)	Blood test for AFP, hCG, andestriol to detect Down syndrome, Trisomy 18, Neural tube defects etc.
quad screen	Not available	INHIBIN A - 2100(appx.)	Blood test (measures AFP, hCG, estriol, and inhibin-A)for Down syndrome, Trisomy 18, NTD etc.
Level 2 scan	100/-	1800(apprx.)	Detects certain birth defects in baby.
NIPT	Not available	25,000(appx.)	it tests small amount of DNA of baby that is naturally found in blood of mother for Down syndrome, trisomy 18, and hemophilia
Diagnostic tests			
CVS	Not available	Karyotyping-11,000(apprx.) FISH - 13,000(appx.)	Sample is taken from chorionic villi of the growing fetus. and sent to laboratory for the test e.g. karyotyping or enzyme analysis for Down syndrome.
Amniocentesis	Not available	Karyotyping-12,000(apprx.) FISH - 11,000(appx.)	Sample is taken from amniotic fluid present around the growing fetus e.g. karyotyping , FISH for trisomy 18.

Ethical issues of prenatal testing

- The option left after prenatal testing is either to continue or terminate the pregnancy.
- Patient must have complete information about the tests done.
- Confidentiality should be respected.
- Informed consent must be taken.
- After confirmation of diagnosis for birth defect whether it is lethal (can cause death) or not ,in India after 20 weeks MTP is not allowed. E.g. in a recent news it was heard that the court declined a woman's plea to abort her 26 week old fetus detected with Down's syndrome. The court denied the plea by saying that the fetus poses no danger to the woman's life. The child with this syndrome is not as seriously handicapped as it to be terminated.

Detection of birth defect after delivery (in child)

Certain birth defects are diagnosed only after the birth of baby. Early neonatal screening plays a important role in this. Sometimes, the birth defect is seen immediately at birth (club foot). In other cases birth defects might not be diagnosed until later in life (heart defect).

Whenever there is a health problem in a child, the doctor/nurse may look for birth defects by taking a medical and family history by asking questions. Doing a physical examination (anomaly detection) and sometimes recommending further tests (biochemical scrreening) may also be beneficial.

If the diagnosis is still uncertain, they might refer the child to a bigger hospital where a specialist in birth defects and genetics can deal with the patient. An exact diagnosis may not be made immediately because it takes time for further testing. Moreover there are many unknown causes which make diagnosis difficult.

For PKU, congenital hypothyroidism, and other inherited metabolic defects, neonatal screening is only option. It allows early dietary intervention for PKU and thyroid replacement therapy for hypothyroidism, both of which are critical to avoid mental retardation. Neonatal diagnosis of congenital hip dislocation provides the best chance for correcting this disorder through noninvasive techniques.

How to Deal with Birth Defects after the Confirmation of Diagnosis

Not all birth defects are permanent or equally debilitating. Some are curable through early treatment, surgery, or therapies. However due to their disabling nature, some birth defects create a need for lifelong medical and supportive care for affected child. Certain conditions require very little additional expense to take proper care of the child.Others may require expensive medical equipment, extended hospitalization or repeated surgeries, or regular medication.This puts a lot of burden on the family.These also create various other social issues, e.g., stigmatization.

The first thing that most of the parent wants to know whether baby in the womb is healthy or not. So when they are told that their baby has birth defect they get shattered.

What to do If birth defect is detected during pregnancy (in fetus)

- It is often difficult to take a decision in such situations. During thiss time couple should support each other and try to relax and feel normal.
- Reliable and accurate information can help parents to take tough decisions well in time. (MTP <20 weeks). e.g. Women who come to know that fetus has thalassemia or Down syndrome may go for termination of pregnancy, if it is detected well in time.
- If gestation age is more than 20 weeks you should continue routine antenatal checkup.
- You must go for institutional delivery (in a hospital) so that newborn can get timely intervenion. e.g. Consider the case of heart problems, where a major artery may be connected incorrectly or a pumping chamber may be missing. Prenatal detection and immediate treatment at birth can prevent the sudden oxygen deprivation and shock at the time of birth. If the doctor is already aware of the finding and he can well manage the things in time.

What to do if birth defect is detected after delivery (in child)

As already discussed early neonatal screening can provide an opportunity to get an early treatment. e.g. pacemaker for a heart problem, surgery for anal atresia etc.

Many birth defects have no specific treatments. But the developmental delays that occur in first 3 years of life can be dealt with a multidisciplinary approach. Often the defects diagnosed in the baby at some later stage of life can cause irreversible life-long mental, physical, auditory or visual disability. e.g. a baby with congenital hearing loss can have delayed speech and language development if diagnosed later in life. Early detection through prenatal testing can help early management.

Early intervention services can include feeding support, assistive technology (tools, devices and aids that make everyday tasks easier for people with disabilities), occupational therapy, physical therapy, speech therapy, nutrition services, and social work services

- *Parents sometimes over-protect their children out of a desire to keep them safe and healthy. However, over-protection can make a child feel that he is incapable of making good decisions. This can have unintended negative consequences as children become adults and need to care for themselves. Parents need to understand that, as much as possible, children with birth defects should be made to feel normal—like any other kid.*

Steps for Management of birth defects

- Careful clinical evaluation of pregnancy.
- Review of family, prenatal history, and perinatal history.
- Laboratory studies: Chromosome, DNA, biochemical assays
- Imaging studies: USG,Echo, X-rays
- Medical management and genetic counseling
- Autopsy and specific pathological analyses if still birth/early neonatal death so that next pregnancy could be managed properly.
- Visit to social worker and support groups for emotional support.

Role of parents

Parents of babies with genetic defects / congenital malformations often find themselves in shock after hearing the news about defect in the baby.

When they look at healthy babies in the world around them they think, "why us?"

There is no answer to this question for them. They feel frustrated. They have to face many problems in their life. Regular visits to the doctor may affect their routine. This may affect their job. They may undergo emotional, psychological and financial stress.

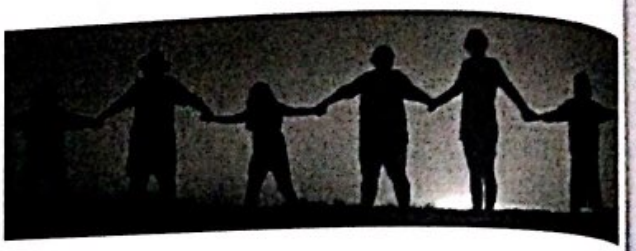
Steps to be followed by parents of child with birth defect

Parents must acknowledge their emotions of grief, anger or shock and talk about them with spouse/partner and other family members. A very heart touching example of a NGO "Sadhna Society" Chandigarh can be quoted here . Dr. Bhawna Tyal Director of the society got inspiration to start this society from her own daughter with Down's syndrome. At present she is taking care of so many special children in her society .

- Get support from healthcare professionals about their queries and find out the support group.
- Let themself enjoy the welcome of their baby with birth defect in the same way any parent would do with a normal health baby.
- Parents are often worried about the next baby. These fears are natural.But instead of worrying parents should go for investigation of the cause of defect so that in next pregnancy this factor could be taken care of.
- Parents should visit a genetic counselor and educate themselves with the knowledge gained after discussion with doctor.
- Parents should seek for early intervention, e.g., aids to make everyday tasks

easier for children with birth defects, occupational therapy, physical therapy, speech therapy, etc.

Role of family



Family undergoes psychological and emotional burdens when they have a child who has a birth defect. There are many factors that can impact family finances such as travelling, medical costs and other health-care related costs as well as specializedchild care management.

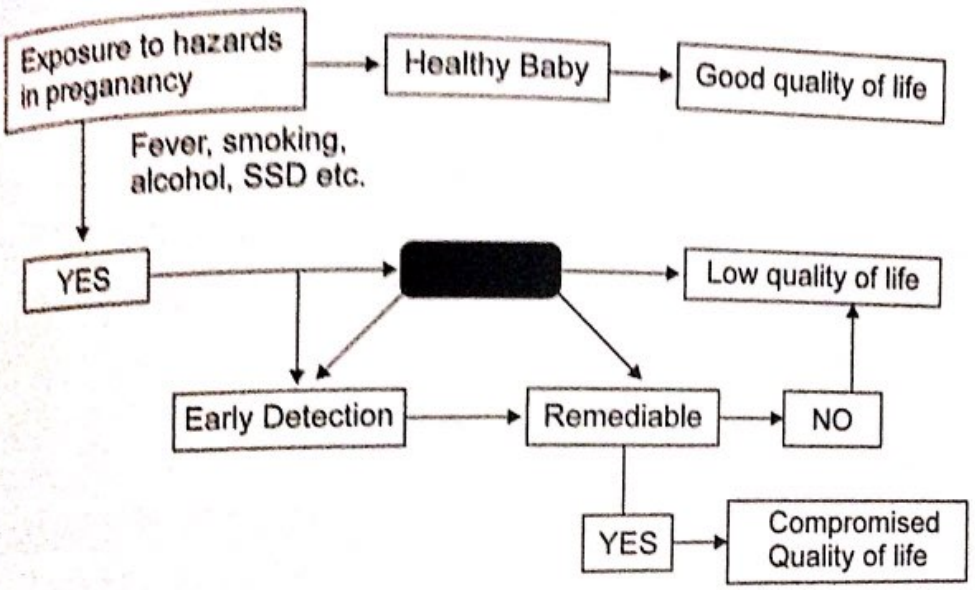
Siblings and other children in the family often feel neglected. These feelings may be justified but result in behavioral issues and depression.

- Families should understand the difficulties parents and child are facing and provide emotional support to them.
- Family members should support parents in every possible way, psychological or financial, to the possible extent.
- Families should encourage parents of such babies for early consultation.They should provide support in follow-up visis if needed.
- They should love the child with birth defect just like a normal baby. It is good if they interact with the baby as much as possible. The family should assist the parents in other household works as parents have to visit hospitals with their child every time
- Family members can take care of siblings when the parents go to the hospital.

Prevention of Birth Defects

"Prevention is better than cure"

birth defects have a serious physical, psychological and financial impact for the family. cause of more than 60% of birth defects is unknown. Yet certain measures can be taken in reduce the risk of their occurrence.



Before pregnancy

- Marriage between very close blood relations like uncle- niece, aunt-nephew, among cousins should be avoided.
- Avoid pregnancies before the age of 18 years and after the age of 35 years.
- Rubella infections are cause of birth defect. Vaccination programs in several, mostly developed, countries prevent virtually all cases of congenital rubella. Giving measles-mumps-rubella (MMR) vaccine to girls prevents congenital rubella syndrome in baby born to women after marriage.
- Optimization of medical disease before planning pregnancy e.g. epilepsy, diabetes. Avoid unnecessary intake of medicines, specially anticonvulsants, anticoagulants, thalidomide and misoprostol (When planning to conceive or during early pregnancy).

➢ Consult a doctor before planning the pregnancy; if

a) There is incidence of birth defects in your family.

b) You have had difficulty in conceiving or have had a series of miscarriages, still births, twins, delivery by operation (Caesarean), obstructed labour/prolonged labour(more than 12 hours) and/or severe bleeding in previous pregnancy.

c) You have Rh-negative blood type.

d) You have diabetes/other chronic disease.

During Pregnancy

- Woman having frequent pregnancies, having a history of miscarriage/ abortion/premature deliveries, must get expert prenatal care
- Proper intake of folic acid and well-balanced and nourishing diet supplemented with green leafy vegetables, proteins and vitamins.
- Rh Negative women should be given Anti D injection at around 28 weeks and 36 weeks of pregnancy to protect their child from hemolytic diseases.
- Avoid unnecessary drugs and medications, smoking, chewing tobacco, consuming alcohol and narcotics, unnecessary exposure to chemicals and radiations (X-rays), exposure to illnesses like measles, mumps, etc. especially during the first 3 months of pregnancy.
- Take precautions against lead poisoning (Lead is found in lead-based paints, including paint on the walls of old houses and toys)
- Prenatal diagnosis for familial disorders by e.g. Ultrasonography and maternal serum screening

can be used to detect serious fetal anomalies, including neural tube defects and chromosomal disorders.

After birth

Early neonatal screening can be implemented to avoid the complications of birth defects.This increases an affected child's possibilities of a better quality of life.

- Infants can be screened for a variety of genetic, hematological, metabolic, and hormonal disorders to provide early diagnosis.
- Early detection and treatment of birth defects can help prevent physical and intellectual disabilities. E.g. congenital hearing impairment can result from a number of environmental or genetic causes. Without early testing, a diagnosis of hearing loss might not occur until a child is two or three years old, resulting in delayed speech and language development.
- Another example is phenylketonuria (PKU), which is an inherited disorder characterized by the inability to synthesize theamino acidtyrosine from phenylalanine. Treatment of this condition involves a diet low in phenylalanine and high in tyrosine. Treatment results are best if this diet is implemented at birth. If this is done the child can lead a relatively normal life.

Treatment Strategies

An important aspect of reducing the impact of birth defects is the access to treatment. Some birth defects cannot be treated cost-effectively, others can be corrected partially or entirely by therapies that are clinically cost-effective.

Treatment depends on the type of defect and the baby's condition.

- Some birth defects can be treated before birth (spina, bifida) but that is rarely available in India.
- Some birth defects can be treated or repaired at the time of birth.(CHD)
- Some birth defects are so small that they don't need to be treated.(periauricular tags,syndactyly)
- Some birth defects can be treated after few years of life.

Home Care

Parents may be guided to follow specific instructions for monitoring, feeding and bathing infants with the birth defects. Although they need specialist guidance for rehabilitation, but to follow the guidelines to practise the child is a hometask for parents too.

Medicines

These may be used to reduce complications that arise due to certain defects.In some cases medication may also be prescribed to mother to correct an abnormality before the birth of baby. Cystic fibrosis would involve a multidisciplinary team (physician, physiotherapist, nutritionist, and social worker) to provide conventional therapy (antibiotics, pancreatic enzyme replacement by ingestion of capsules with meals, and nutritional support) and treatment for associated complications. Another example is of use of biotin in high doses for treatment of biotinidase deficiency.

Surgery

Surgery is also an option to treat many birth defects, e.g., cleft clip or cleft palate. Prenatal heart and renal tract defects are often treated surgically while the child is still unborn. Although these are not used on a wide scale in India. Operation of affected organ is also done after birth of the baby. This reduces harmful symptoms in children who have some physical birth defect (Plastic surgery for either cosmetic or health benefit).

Rehabilitation of Children with Birth Defect

Many infants with severe birth defects experience lifelong disability requiring long-term treatment or rehabilitation. They may have serious lifelong consequences for both the patient and the family. Those affected may require psychological, emotional, and social support on a continuing basis; this support should include referral to and assistance from social services such as education and social welfare. e.g., a child born with Down's syndrome has low IQ level. A child may need physical, occupational, and speech therapy for his or her development.

In many cases, appropriate education and rehabilitation for children and adults substantially increases their ability to function independently and contribute to family and community responsibilities. It also helps to overcome the isolation and stigmatization experienced by people with disabilities and encourages those with disabilities to live lives that are as normal as possible and are integrated into society.

Early screening & intervention focuses on helping eligible babies and toddlers learn the basic and brand-new skills that typically develop during the first three years of life

- *physical* (reaching, rolling, crawling, and walking)
- *cognitive* (thinking, learning, solving problems)
- *communication* (talking, listening, understanding)
- *social/emotional* (playing, feeling secure and happy)
- *Self-help* (eating, dressing).

Child may need different rehabilitation services depending upon the type of birth defect.	
• Assistive technology (devices a child might need) • Audiology or hearing services • Speech and language services • Counseling and training for a family • Medical services	• Nursing services • Nutrition services • Occupational therapy • Physical therapy • Psychological services

A successful story of a NGO in India helping special children

The occurrence of a shocking incident sometimes makes you stronger and inspires you for Noble task. As it is true of "Sadhna Society" for mentally handicapped where overwhelmed with the deep feelings for their own Children, few parents of children with intellectual disability got self-motivated to serve similar children in society and joined their hands to conceive Sadhna Society for the mentally handicapped.For the purpose to educate and train the special children Sadhana society started their **Institute 'Sadhana vocational training institute' for the mentally handicapped in Chandigarh**. The institute opened up new avenues for intellectually disabled under the expert guidance of educators, physiotherapist, speech therapist, yoga and dance teachers and the parents working voluntarily hard. Children are put to various schemes and programmes with the scientific approach apart from the basic academic.

Our students do us proud in Olympics every year by winning in different sports at zonal and national levels. They have brought us proud by winning awards in music and dance competitions Tejaswani Sharma the Music Prodigy is the star of our Institute. Not only intellectually disabled but also suffering from cerebral palsy and visual problems is star or miracle of god blessed with the rare gift of singing. Restricted to the age of 8, unable to walk, talk or even blink her eyes she struggled for life since her birth, but it is music that revived Tejaswani when she was 11. Her mother during the car drive suddenly noticed her humming the tune been played on the car stereo that was a momentus occasion for the family.To encourage her talent she was enrolled in a Music Academy where she learnt number of bhajans, ghazals and classical and folk song. Thereon it was her continuous journey towards her goal to be a professional singer, when she grows up.And today, her dreams have come true she has become a world famous singer apart from winning a number of music competition she got many awards.

Sadhna Society

Tejaswini sharma singing performance

Dr. Bhawna Tyal, Director, Sadhna society NGO, Chnadigarh

Treatment Options Available for Specific Birth Defects

Talipes or clubfoot

- Early intervention is critical to correcting the condition and obtaining a foot that is supple, painless, plantigrade, and of normal shape and function.
- Deformity can be resolved through manipulation and passive stretching,
- By 12 weeks of age, the soft tissues of the infant foot are far less pliable, so only surgery can correct the condition.
- More severe cases require treatments like plaster casts, splints, special shoes, and sometimes surgery followed by exercises
- Baby should be encouraged to promote normal growth and development, including sitting up and crawling, while the foot is immobilized.

Cleft lip and/or cleft Palate

- Prenatal detection of the condition, available in some urban areas, can help parents to prepare for feeding and other special care of their child.
- Support and education are key components of ongoing care.
- Surgical repair of the lip can be done at 3 months and palate repair at 6 months of age.
- Comprehensive treatment can include speech and language therapy, preventive and restorative dental care, orthodontics, secondary surgery, otolaryngology for hearing problems, and psychological counseling.

Congenital heart disease (CHD)

- Depending on severity, these are typically treated with surgery, drugs, or a mechanical aid like a pacemaker.
- For example, medicines can control an irregular heartbeat, coarctation of the aorta, or tetralogy of Fallot.
- Heart catheterization, where the doctor threads a thin, flexible tube called a catheter through a blood vessel in the groin, may also be used to repair a defect. Others may be helped by surgery e.g. closing holes in the heart with stitches or a patch, repair or replace heart valves, widen arteries or openings to heart valves, repair of complex defects.

Developmental dysplasia of the hip

- Simple, noninvasive postural treatments can prevent a severe and crippling condition.
- This requires a series of complicated surgeries and rehabilitations. Treatment appears to be equally effective at 6 weeks and can be undertaken successfully in children up to 2 years age.
- Early treatment maintains the thighs in a flexed and partly abducted position so that the head of each femur remains deep within the acetabulum, encouraging it to encompass the femoral head more completely. To maintain this position, the thighs are splinted for weeks to months (up to a year).

Spina Bifida

- Severe cases require surgery within 48 hours of birth, followed by special exercises.
- The child may still need to use leg braces and crutches.

Missing limbs

- Parents are referred to an orthopedic specialist who helps fit the child with a prosthesis as early as possible.
- Intense physical therapy follows to help him or her learn to use it.

Down Syndrome

- As such there is no treatment for this syndrome, as it cannot be "cured," but medical therapies may be necessary for accompanying visual or hearing impairments.
- In addition, early intervention therapies like speech therapy and physical therapy help advance development.

Thalassemia

- Child requires regular blood transfusions combined with iron chelating therapy. This regime allows many people with the disorder to survive into their twenties and thirties.
- A more expensive and higher-risk option is stem cell transplant from bone marrow or cord blood. This can cure the disorder but is not affordable in low-income populations.
- Without diagnosis and treatment, patients with β-thalassemia major usually die early in childhood.

Glucose-6-phosphate dehydrogenase deficiency

- Interventions focus on reducing the occurrence and severity of hemolytic crises. These episodes can be precipitated by triggers for hemolysis such as infections such as hepatitis or pneumonia; exposure to certain chemicals and oxidative medications, including some antimalarial drugs; and consumption of fava beans.
- In hemolytic crisis a blood transfusion may be required.

Cystic fibrosis

- An early diagnosis is the key for prevention of CF since many high-risk couples have a second affected child before the diagnosis has been established for the first child. Ideally,
- Treatment of CF would involve a multi disciplinary team (physician, physiotherapist, nutritionist, and social worker) to provide conventional therapy (antibiotics, pancreatic enzyme replacement by ingestion of capsules with meals, and nutritional support)

Phenylketonuria (PKU)

- Increased phenylalanine levels would be toxic to the fetus and cause severe mental retardation, birth defects, heart disease, and low birth weight.
- The mainstay of treatment is a phenylalanine-restricted diet.
- By limiting daily consumption to only 250–500 milligrams of phenylalanine per day, a positive nitrogen balance and safe plasma levels of phenylalanine can be maintained.
- Reduced phenylalanine levels improves IQ and neuropsychological outcome.

Hemophilia A and B

- A series of exercises that facilitate absorption of the hematoma to improve joint range and strengthen muscles, which reduces the frequency of bleedingcan be taught to the patient.
- Home care programs can be organized so that patients can administer their own Factor VIII infusions with the onset of symptoms.

गीतांजलि के सर्जन ने की 700 ग्राम वजनी नवजात की हार्ट सर्जरी

उदयपुर। गीतांजली हॉस्पिटल के कार्डियक थोरेसिक एवं वेसक्यूलर सर्जन डॉ संजय गांधी ने जयपुर के एक निजी हॉस्पिटल में 24 सप्ताह में जन्मे 700 ग्राम के नवजात की जटिल हृदय सर्जरी सफलतापूर्वक सम्पन्न कर नवजात को नया जीवन प्रदान किया। यह राज्य में ऐसा दूसरा सफल ऑपरेशन है जो जयपुर एनआईसीयू में किया गया है इससे पूर्व गीतांजली हॉस्पिटल में ऐसा ऑपरेशन हुआ था। जयपुर निवासी नेहा माथुर ने जयपुर के कोकून हॉस्पिटल में 700 ग्राम वजनी नवजात को जन्म दिया। नवजात के सांस लेने की परेशानी के चलते उसे एनआईसीयू में वेंटीलेटर पर लिया गया था। इको-कार्डियोग्राफी में पता चला कि हृदय की दो मुख्य धमनियां आपस में जुड़ी हैं जिस कारण उसे वेंटीलेटर से हटाना संभव नहीं हो पा रहा। नवजात की नाजुक हालत को ध्यान में रखते हुए गीतांजली हॉस्पिटल के प्रशासन ने

अपनी कार्डिक टीम को जयपुर भेजने का निर्णय लिया जिसके तहत डॉ संजय गांधी, डॉ अंकुर गांधी एवं समस्त ... कोकून हॉस्पिटल पहुंचे और ... में ही ऑपरेशन कर हृदय की जुड़ी हुई धमनियों को ... किया। नवजात अब स्वस्थ है।

First separated New Year for conjoined twins

City doctors successfully perform genital reconstructive surgery

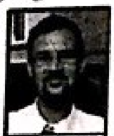
By Vikas Vaidya

BIRTH of a child in a family usually brings a delightful shower of happy moments. It was no different for a family from Nagpur when a girl child was born. As time passed by, the world of their little girl was filled with beautiful dresses and dolls and trinkets. But then one day, the reality hit the family like a bolt from the blue. Their little girl, now 12, was, in fact, a boy!

It all started 12 years ago when Gopi (name changed) was born in a well-to-do family. She was developing normally and was a healthy child. She got good education and was also extraordinary in sports. The only concern of parents had been a swelling in her genital region and some atypical appearance. They discussed this with their doctor but were always reassured.

At the age of 12 years, Gopi started growing tall and muscular. Her voice was becoming a bit masculine and so was her behaviour. Her physical development is very different from girls her age. All this left Gopi's parents obviously panicky. They consulted Dr Hari Mangtani, Pediatric Endocrinologist of the city. He referred the case to Dr Dinesh Sarda, well-known Pediatric surgeon and Pediatric Urologist. After a complete work up what came to light shocked the parents to the core. It turned out that Gopi is, in fact, a male child internally with 46XY chromosomes.

Dr Hari Mangtani



Dr Dinesh Sarda

The swelling in the genital region, that the parents had discussed with the doctor then, was in fact one of the testes.

Once the diagnosis was confirmed, it was thoroughly discussed with the parents and it was decided to give Gopi her real identity as a boy. Pediatric Pschycologist Dr Manju Giri came into the secene and had a tough task making the child understand what had happened and how to embrace the change. Meanwhile, the most challenging part for the doctors now was to make the genitalia appear as that of a male. This reconstructive surgical challenge was taken up by Dr Dinesh Sarda at his Ketki Nursing Home, Dhantoli. Presently Gopi is undergoing psychosocial and behavioral training to face the world afresh as a boy. The family members are happy that even if it took so many years, the psychological ordeal that Gopi would have had to face lifelong potraying what he is not, is now over.

(Contd on page 3)

5-year-old 'butterfly' boy at PGI for treatment

Chandigarh: A five-year-old suffering from dystrophic Epidermolysis Bullosa (EB), a painful and rare genetic skin disease, is being treated at PGI. Kids with EB are often referred to as 'butterfly children'.

The kid has a new genetic alteration, which caused the rare skin disease. A collaborative research between the Institute of Genomics and Integrative Biology (IGIB), New Delhi, and PGI has been able to uncover some novel variants in India causing EB. Consequently, molecular diagnosis and prenatal screening have recently helped one such case to terminate the pregnancy as the foetus was found to have a genetic mutation for the rare genetic condition, which makes the skin extremely fragile, almost like a butterfly's wing. The 'butterfly' kids are prone to blisters.

In the area of genetic skin diseases, IGIB has been collaborating with the department of dermatology, PGI, and has been able to solve a number of cases of such diseases with molecular diagnosis. PGI has 40 Indians affected by this rare skin disease.

In this rare condition, the skin becomes fragile and breaks down in case of any minor pressure or trauma. EB is caused due to mutations, which affect the proteins involved in the architecture and anchoring of the skin layers. The disease has multiple levels of severity from mild to moderately severe i.e. dystrophic. People with severe form EB can ultimately die.

"In the recent paper, we described the utility of genomics approach (whole exome sequencing). This approach makes it cost-effective, time-effective ...

Rarest of the rare

There are 7,000 documented rare diseases in India with an estimated over 80% of them attributed to genetic causes. It is widely believed that around 70 million Indians suffer from genetic diseases.

PAINFUL DISEASE

- Epidermolysis Bullosa (EB) is a rare genetic skin condition
- The skin becomes extremely fragile and forms blisters from minor friction or trauma
- Kids suffering from this disease are often referred to as Butterfly children because their skin is as fragile as a butterfly wings
- Different forms of EB exist depending on where the blisters occur within the different skin layers

Recent advances in treatment (available surgeries)

Some Case Studies

Case study 1.

A woman name A wife of B resident of Chandigarh visited genetic counselling center. She was referred from OBG, OPD. Parents already have a baby with Down Syndrome. Her age is 30 years.

She has been booked at PGI for her antenatal care. She is gravida 3. In her first pregnancy in 2012 she had delivered a baby through emergency Caesarean due to eclampsia. A female baby was born. Baby did not cry at the time of birth. Baby was diagnosed to have down syndrome at a later stage. Mother was told that her baby had low IQ. Baby expired at two years due to frequent seizures.

In 2015, by her second pregnancy she delivered a male baby through normal vaginal delivery. That baby is normal and is of 3 years now and living a healthy life.

For her current pregnancy she reported at PGI at 14 week. She had not taken periconceptional folic acid. Her family history was normal. There was no history of consanguinity of marriage. Her personal history indicated she had increased thyroxin level. She was on medication for this. However her other investigations of blood were also normal (Hb, HIV, HCV).

She was was advised for Amniosentesis and level 2 scan at 16 week. Fortunately test reports came out to be normal. she continued her pregnancy and delivered a healthy female baby.

Case Study 2.

Another mother named M aged 27 years wife of N resident of Roop Nagar, Punjab visited genetic counselling center for preconceptional counselling. In her first pregnancy she delivered a male baby. Baby expired at 4 and half years due to neuroblastoma. Baby was undergoing treatment from PGI Chandigarh. In her second pregnancy she had gone for MTP by medicine as her first baby was ill. Her family was not able to afford her second pregnancy. There was no family history of birth defect. She was told that there may be some

risk in next pregnancy too.

At her visit to centre both mother and father were advised to go for karyotyping. Reports came out to be normal. Mother was advised to start folic acid and take it for atleast 3 months before planning her next pregnancy. She was advised for early booking in her next pregnancy and go for all recommended tests as advised. Specailly a level 2 USG at 18 week in next pregnancy. If any sort of tumor is detected in next pregnancy, then pregnancy can be terminated with MTP. Also after delivery they should go for ultrasonography of baby after every six months until baby achieves an age of one year. For now parents do not need to undergo any further test.

Case Study 3.

Mother S age 33 years wife of T village Roop Nagar visited genetic counselling OPD. In her first pregnancy, she had an abortion, at eight month of pregnancy. In her second pregnancy she delivered a male baby through normal vaginal delivery. This baby is of eight years and living a healthy normal life. In her third pregnancy she delivered a male baby by normal vaginal delivery but baby died after four days due to Glutaric aciduria type-1. She told that baby had stopped taking milk after two days of birth. Baby had seizures. Baby had difficulty in breathing and baby used to remain irritable. In her current pregnancy her POG is 19 week. There is no history of consanguinity of marriage. She has increased thyroid level. She had taken folic acid, iron and calcium during her current pregnancy. There was no history of preconception folic acid intake. Her ultrasonography report is normal. She has been referred for genetic counselling in view of her previous baby that got expired due to blue glutaric aciduria type-1.

No DNA testing was done in expired child. No karyotyping was done in parents too . But now as it is 19th week pregnancy, she is late for the test because report will take few weeks to come and if there is any problem in the report, it will be very late for doing MTP. As MTP is to be done till 20th week of preganncy. If she would have reported earlier at approximately sixth week of pregnancy required tests would have been conducted in time.

Although she was advised for some required tests but,she could only continue her current pregnancy. She delivered a male baby at PGI. Baby had normal morphological feaures at the time of birth. After after a few days baby stopped feeding. Baby was investigated for DNA testing. Again this baby too reported to have Glutaric aciduria type-1. Her early booking at PGI could have helped her for early diagnosis and timely intervention for MTP.

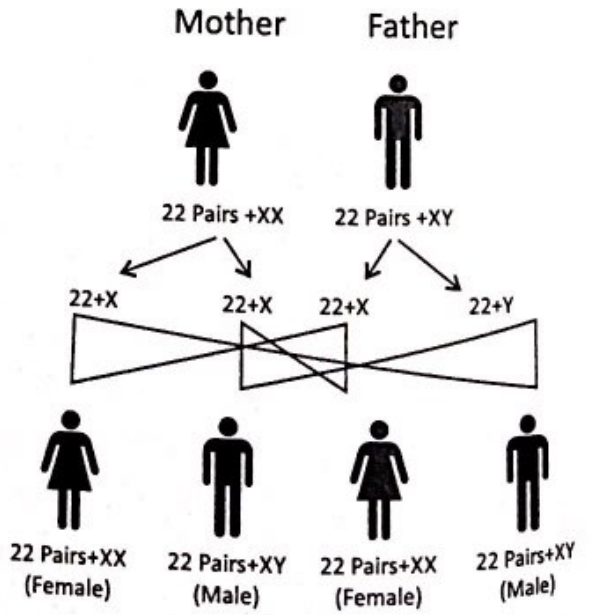
Case Study 4.

A female aged 33 years (Name P w/o Q, Punjab) visited genetic counseling room no 2036 ,PGI in March 2017. She was referred through General Gynaecology OPD to genetic counseling room i/v/o previous baby having congenital disorder. As per her obstetric history she was G3 P201. She is a known case of secondary infertility and oligomenorrhea. She is taking treatment for infertility. Her previous baby has epidermolysis bullosa. First baby (female wt- 2.6 kg) got delivered through cesarean section at a private nursing home in Ropar. The baby had skin lesion on face and foot. Baby was referred to skin department of PGIMER; Chandigarh on the day of delivery. Epidermolysis bullosa was diagnosed in the baby, by the doctors. Doctors advised her not to go for any test till baby is 45 days old. KMNO4 compressions were advised. However baby died after 1 week. Mother of the baby had itching all over the body during pregnancy. Now the patient is planning for her next pregnancy and has come to PGI for infertility treatment. After her checkup she has been referred to dermatology department as there is some skin lesion in perineum. She has been advised for some routine investigations, i.e., blood group/TSH/FSH/LH/GTT/Montoux. She has been advised to start folic acid and tab fluconozole along with vaginal pessary.

As no autopsy report of the previous baby or any blood sample is available parents have been advised to go for karyotyping by genetic counseling in Room 2036. After the karyotyping it will be seen whether there is some affected gene present in the parents by DNA testing , the same will be tested in next baby also. But parents were not willing as they said they cant afford the test. so they were advised to continue folic acid atleast 3 months before pregancy as well as to continue even after conception. She has been advised for early booking in next pregancy.

Understanding the Mechanism- How and Why Genetic Disorders Occur?

- Each organ of human body is made up of cells. Each normal cell contains 46 chromosomes(23 pairs).
- Normal cell - 22 pair of chromosomes + XX (female), 22 pairs of chromosomes + XY (male)
- Gamete cells (ovum and sperm) only contain 23 chromosomes (rod like structures present in the nucleus of cell , made up of DNA). Each chromosome contains thousands of genes.
- When the genes and chromosomes are 'normal ' a healthy child is likely to be born. If there is some defect in these chromosomes, congenital disorder or genetic disorder may occur.
- During intercourse husband's sperm (22 chromosomes + X or 22 chromosomes + Y) and wife's ovum (22 chromosomes + X or 22 chromosomes + X) meet.
- These two gametes merge into one zygote cell. The new zygote contains 23 chromosomes from father and 23 chromosomes from mother. Thus 46 chromosomes are attained in each cell.
- Being male or female depends on whether we have XX or XY sex chromosomes as 23rd pair.



In newborn incidence of chromosomal anomalies is 5.6 per 1000 live births. They are of 2 types.

Numerical abnormalities–in Such cases whole chromosomes are either missing from normal sequence of 46 chromosome or is extra. For example Trisomy (three copies of a chromosome) instead of monosomy (single copy of a chromosome). Down's syndrome is probably the most well-known example. Besides trisomy 21, the major chromosomal aneuploidies seen in live-born babies are: trisomy 18, trisomy 13.

Structural abnormalities- when part of an individual chromosome is missing, is extra, switched to another chromosome, or turned upside down. Examples of structural chromosomal abnormalities include *cri du chat* or "cat cry" syndrome. Children with this syndrome have an abnormally developed larynx that makes their cry sound like the mewing of a cat in distress. They also have a small head, misshapen ears, and a rounded face, as well as other systemic defects. These children usually die in infancy. *Cri du chat* is caused by a deletion of a segment of DNA in chromosome no. 5.

Some Basic terms used in the context of genetics

- **Genotype** : (Genetic constitution) -Tt
- **Phenotype:**(outward expression)- tall
- **Homozygous**: (when genes of a pair are alike)- TT or tt
- **Heterozygous**: (when genes of a pair are different)- Bb or Tt
- **Dominant gene:**In this the phenotype effect comes in both homo(TT) and heterozygous (Tt), i.e., both combinations will make a tall child
- **Recessive gene:**In this effect comes only in homozygous (tt)- Short height. In Tt the effect of T will dominate over t = Tall Child
- **Autosomal:** For abnormalities in non-sex chromosomes. Tt (Height character is transmitted through autosome)
- **Sex linked:** $X^{\circ}Y/XX^{\circ}$- If the disease trait is present in X sex chromosome it will be known as X sex linked and if it is present in Y chromosome it will be known as Y sex linked.
- **Case:**Presence of disease trait in one of the chromosome will make the male a case for that disease ($X^{\circ}Y$), since disease causing X° will dominate over normal Y)

e.g. hemophilia

- **Carrier:** As in a female there are two XX sex chromosomes. If there is Presence of a trait of disease in one X chromosome. Female will be carrier for that disease. Symptoms for that disease will not be present in the female ($X^{\circ}X$).Since normal x will dominate over $X^{*}X^{\circ}$).

A clear example of the relationship between genotype and phenotype exists in cases where there are dominant and recessive alleles for a particular trait. This can be understood by a simple example.

A capital "T" is used to represent a dominant allele at a particular locus coding for "tallness" the lower case "t" is used to represent the recessive allele coding for "small height". Using this notation, a diploid plant will possess one of three genotypes: TT, Tt, or tt (the variation tT is identical to Tt). Although there are three different genotypes, but human will be either tall or short (two phenotypes). Human with a "TT" or "Tt" genotype are observed to be tall (phenotypically tall). Only those that carry the "tt" genotype will be observed to be short (phenotypically short).**Figure -1**

Another example may be of famous Bollywood star Amitabh and actress Jaya.(1st generation kids will be tall).**Figure -2**

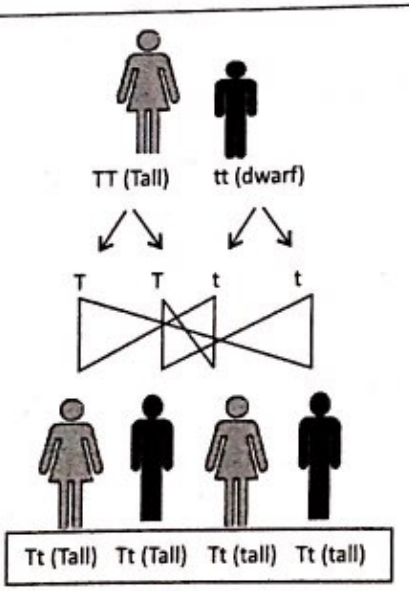


Figure 1

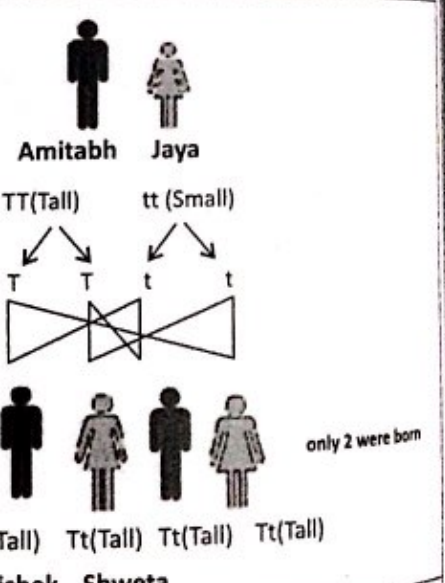


Figure 2

It can also be understood by another simple example for black colour of eyes.As it is known

genes are always present in pairs.If mother and father both have black colour eyes (BB) all the babies will have black colour eyes. If mother and father have blue colour eyes (bb) all the babies have blue (bb) colour eyes.

To represent black colour of eyes we take B alphabet .so genotype will be BB. As B is dominant it will produce black colour eyes in BB and Bb pair. If it would have been recessive it would manifest only in bb.

In the first case we take black colour eye female (BB) dominant and blue colour eye male (bb) recessive. When we make a cross between them. We get the following results in the first generation kids.Bb is heterozygous and B is dominant and is present in all the four outcomes. So all babies with black colour eyes are produced.**(Figure 3)**

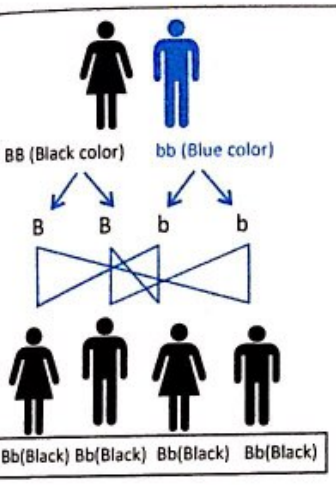


Figure 3

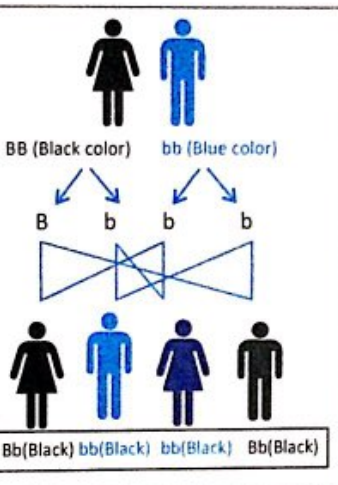


Figure 4

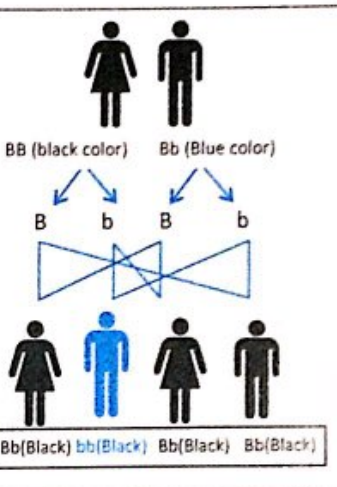


Figure 5

In the second case we consider female having black colour eye (Bb) and male with blue colour eyes (bb). Now in this case the following outcomes will be produced**(Figure 4)**.We can see there are two Bb and two bb. Bb is heterozygous and B is dominant so they will have black colour eyes. Whereas bb is recessive and they will have blue colour eyes.

In the third case we consider female (Bb) black eyes and male also (Bb) black eyes. In this case one BB (black eyes) two Bb (black eyes) and one bb (blue eyes) are produced. There are chances of 3 babies with black eyes and one baby with blue eyes.**(Figure 5)**

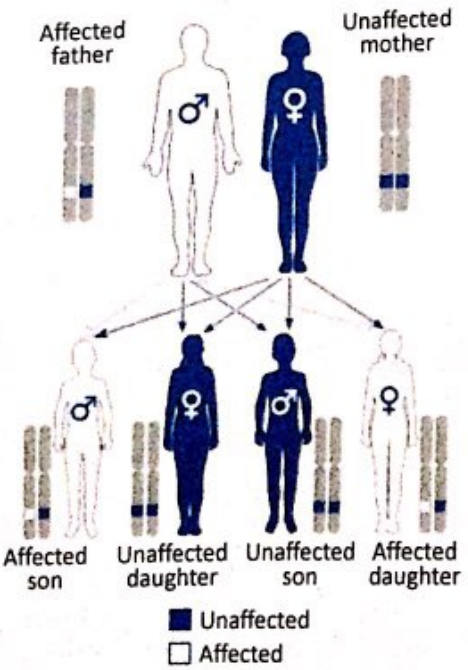
Patterns of inheritance

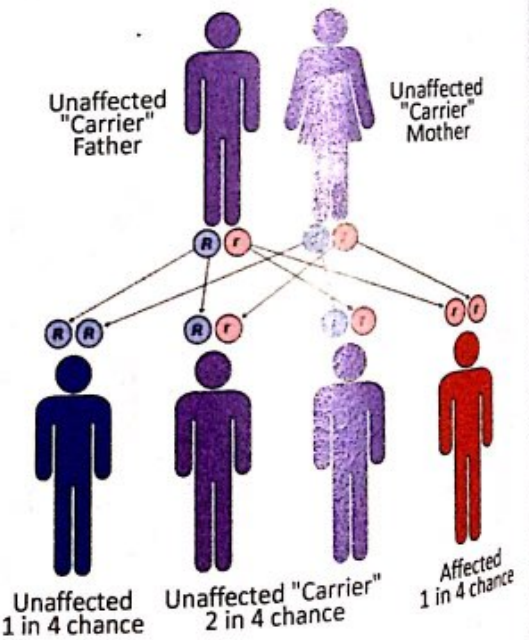
A) Related to sex chromosome.

Autosomal dominant inheritance-When a single gene is present and expresses itself without regard to the other gene at that locus. It is said to be dominant.More than 700 such diseases are reported,e.g.,retinoblastoma, neurofibromatosis, Marfan syndrome achondroplasia, Huntington's chorea etc.

- Each generation shows affected member (no generation is skipped).
- Unaffected individuals do not transmit the trait to their offspring.
- each of their offspring having a 50% chance of being affected.
- Both sexes have equal chances of being affected.

This can be understood by the following diagram for a common autosomal dominant disease, .e.g. achondroplasia. Father is affected by the disease (one affected gene and another unaffected) whereas mother is not. There are equal chances of delivery of 50% male and 50% females.. In one male child affected gene may go and in another unaffected gene may go. Now in the male child that gets affected gene, disease will occur whereas the male child that gets unaffected gene, disease will not occur. It is to be noted here that in both male child the gene that comes from mother is normal. Now the female that gets affected gene from father becomes carrier for disease. It means symptoms will not express in the female child. However, symptoms may express in the next generations.





Autosomal recessive inheritance - When individual with one gene for the trait (carrier) does not express the clinical features. This is found more in cansanguineous marriages.

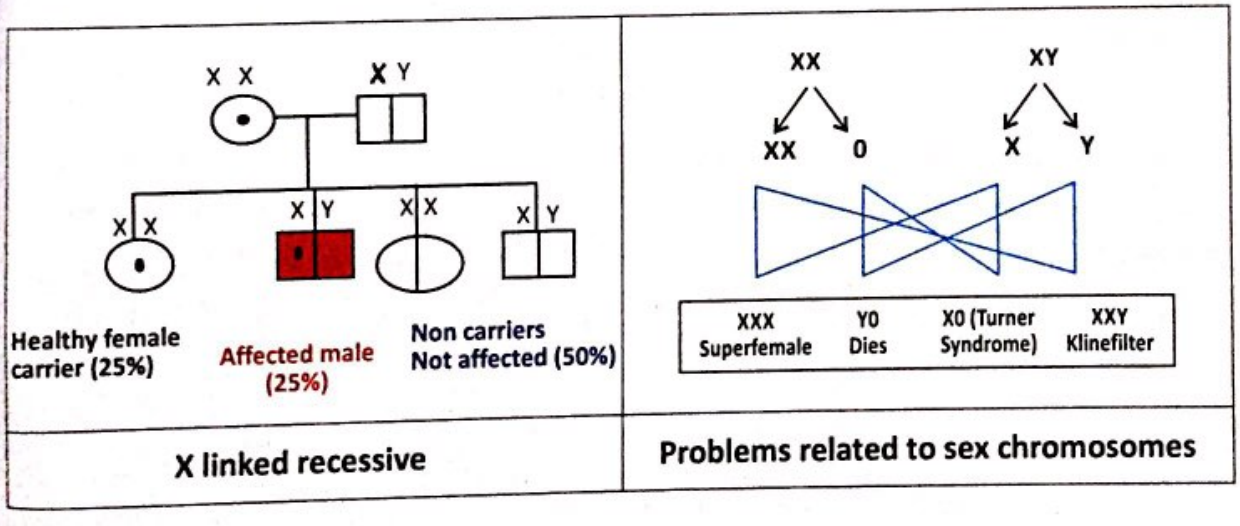
- Only an individual who gets two genes for the trait has clinical manifestation.
- On an average 25% of the offsprings of two carrier parents are affected.
- Both sexes have an equal chance of being affected.

X linked recessive inheritance- The genes involved are located on X chromosome when the female carries a sex-linked recessive trait, she is a carrier. She does not exhibit clinical symptoms. This is because this gene is recessive to the normal alleles located on her second X chromosome.

However since male has only one X chromosomes. Therefore, only a single gene for a particular sex linked traits. All males who carry the defective genes will be affected.

- With rarer exceptions only males are affected.
- While the disease may appear to skip one or more generation, affected males are related through carrier females.
- One half the sons of a carrier female will be affected

e.g. Hemophilia acquired the name the royal disease due to the high number of descendants of Queen Victoria afflicted by it. This is assumed that a mutation occured in the sperm of the Queen's father, Edward Augustus, the Duke of Kent.



X linked recessive | **Problems related to sex chromosomes**

If an affected male marries a healthy female carrier then there is 25% chance – affected female, 25% chance -affected male , 25% chance-unaffected healthy male and 25% chance

– carrier healthy female. If he marries a normal healthy female 50%- carrier healthy female and 50% chance-normal healthy male

B) Related to sex chromosome.

Klinefelter's syndrome- Person with this syndrome are abnormal male (XXY/XXXY). They have nonfunctional testis. Spermatozoa are absent in their ejaculations, growth of hairs on face, pubes, axillae is scanty. They can have gynaecomastia & mental retardation. Incidence 1/1000 male

XYY Syndrome- Person is aggressive, antisocial, often criminal male. Main features are exceptional height (more than 6 feet), and personality/behavior disorder

Turner Syndrome- This is the most common chromosome disorder in humans . its incidence is 1 in 7500 live born girls. They have 45 chromosomes (Single X) instead of XX. They have short stature, infertility, primary amenorrhea; other features include, coarctation of aorta, pulmonary stenosis, and renal malformation. Persons suffering from this syndrome are apparent females with underdeveloped sex glands.

Super females (XXX/XXXX) They have underdeveloped external genitalia, uterus and vagina, and can be tall.

Table- List of chromosomal disorders

Autosomal dominant Achondoplasia,Huntington Chorea Neurofibromatosis, Polyposis Coli Brachydactyly, Marfan Syndrome Retinoblastoma	**Autosomal recessive** Fibrocystic Disease Of Pancreas Phenylketonuria, Albinism, Thalassemia, Color Blindness, Microphthalmos, Infantile Amaurotic Idiocy, Laurence-moon-Biedl-syndrome
X linked dominant Rickets-vitamin D resistant Hypophosphatemia, Blood group	**Sex linked recessive** Hemophilia, Agamma globulinemia Muscular dystrophy, Colour blindness-red green, G6PD Deficiency

Medical Council of India sponsored CME on "A capacity building seminar for strengthening communication skills of doctors for counseling of parents regarding birth defects"

The seminar was organized with the objective of spreading awareness on birth defects. Birth defects are common, critical and costly conditions. They have a high impact in almost all spheres of our life like physical performance, competence and capability in case of major limb anomalies, central nervous system and cardiac anomalies. In some cases it makes a person dependent on others for whole life e.g. absence of limbs. Genetic counseling about various prenatal testing techniques helps a patient to know about possible risk and indication of the test. These tests give a clear and more accurate diagnosis at an earlier stage of pregnancy and parents get more time to seek proper advice.

H.T. CHANDIGARH

PGIMER holds capacity-building seminar for docs

CHANDIGARH: A capacity building seminar for strengthening communication skills of doctors for the counselling of parents regarding birth defects in kids was organised as part of a medical education programme, by department of obstetrics and gynaecology, PGI School of Public Health, Chandigarh, on Thursday.

The programme was sponsored by the Medical Council of India. It was organised for spreading awareness on birth defects. "They have a high impact in all spheres of our life such as physical performance, competence and capability in case of major limb anomalies, central nervous system and cardiac anomalies," said one of the doctors.

It was addressed during the seminar that genetic counselling about various prenatal testing techniques helps a patient know about possible risk. These tests give an accurate diagnosis at earlier stage of pregnancy. Additional professor Dr Neelam Aggarwal was the organising secretary of the seminar HTC

Dance performances by special children

Against this backdrop CMEentitled "A capacity building seminar for strengthening communication skills of doctors for counseling of parents regarding birth defects" was organized was organized to bring together various stakeholders on 8th February, 2018" at Auditorium-Advanced Eye Centre, 2-5 pm.

Dr.Neelam Aggarwal (Additional Professor, OBG Department, PGIMER) was the Organising Secretary to this Seminar. Dr. Amarjeet Singh (Professor, Department of Community Medicine) and Dr.Alka (Research Scholar, Centre for Public Health) were Convener and Co-convener to the seminar respectively.

Experts from various fields' i.e. Gynaecology, Pediatrics, community medicine, Nursing Institutes, Panjab University, Public health department participated in the seminar. Mrs. BhwanaTyal, Director, Sadhna Society vocational training institute for mentally handicapped shared her experiences with Special Children. This was followed by performances by special children. Lecture of expert speakers was followed by interactive discussion with the audience. This initiative will help work towards removing barriers and making existing health care systems more inclusive and accessible to people having babies with birth defect.

This seminar highlighted the needs of birth defect babies. Opinions of target group i.e. parents of birth defect babies will help identify gaps and priorities to reduce health inequalities and plan improvement for access and inclusion. This seminar will act as an advocacy mechanism for the needs of patients through discussion among experts.

Genetic counseling- where available

1. PGIMER, Chandigarh- Genetic clinic and prenatal counseling centre
2. AIIMS, New Delhi- Genetic Unit, Department of pediatrics
3. SGRH,Delhi- centre for medical genetics
4. SGPGIMS Lucknow- Department of genetics
5. Kasturba Hospital Manipal
6. Osmania university, Hyderabad
7. Nizam, Hyderabad
8. Centre for human genetics, Banglore
9. KEM Hospital,Mumbai
10. Koumundi godbole, Pune

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Occurrence of birth defect is an important public health issue. They cause a significant but un- recognized burden to mortality and disability among infants and children. Inadequate knowledge about this issue among parents could result in delayed intervention in their affected child as well as delayed detection and decision making in next pregnancy. Children who survive and live with birth defect face life long physical, behavioural, social challenges. Hence there is a need to increase awareness among parents of such babies. This Booklet intends to give knowledge regarding birth defects.

The authors

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