

**A REVIEW OF CONGENITAL DISORDERS IN NEONATES****\*Dr. Madhuri Mahadar<sup>1</sup>, Dr. Deepa Khajgiwala<sup>2</sup>, Dr. Lad Meenal<sup>3</sup>**

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**INTRODUCTION**

A Congenital Disorder is a medical condition that is present at birth. It can be detected before birth, at birth or even years later. Congenital anomalies are an important cause of childhood mortality and morbidity. Congenital disorder is condition that exists at birth and then develops through the rest of one's life. A congenital malformation is a congenital physical anomaly that is structural defect which is perceived as a problem. Birth defect is a widely used term for a congenital malformation i.e. a congenital, physical anomaly which is recognisable a birth.

Congenital malformations can be grossly divide in following types-

- 1) Limb malformations i.e. Dysmelia, Skeletal defects
- 2) Defects of Nervous system
- 3) Defects of Cardiovascular system
- 4) Defects of Gastrointestinal system
- 5) Defects of Genitourinary system
- 6) Genetic disorders
- 7) Congenital infections
- 8) Congenital metabolic diseases.

Limb malformations include all forms of malformations of limbs such as Amelia, Electrodactaly, Phacomelia, Polydactaly, Syndactaly, and Oligodactaly, Cleidocranial dystosis.

Congenital malformations of nervous system include neural tube defect such as Spina-bifida, Meningocele, Meningomyelocele, Hydrocephalus, and Anencephaly.

Congenital malformation of Cardiovascular system include heart with Patent ductus arteriosus, Atrial septal defect, Ventricular septal defect and Fallots tetralogy, Acyanotic defect, Coarctation of Aorta, Cyanotic defects, Transposition of great arteries, Persistent Foetal circulation.

Defects of Gastrointestinal system such as Congenital pyloric stenosis, Cleft lip and palate, Hirsh sprung's disease, Imperforate anus, Umbilical Hernia.

Defects of genitourinary system are Bladder exstrophy, bifid ureter, Agenesis of kidney.

Genetic disorders which may be single gene defect or multiple gene defects.

Single gene defect may arise from abnormalities of both copies of an autosomal gene (a recessive disorder) or of only one of two copies (a dominant disorder). Some conditions result from deletion or abnormalities of few genes located contiguously on a chromosome e.g. Klienefelter's Syndrome, Mongolism, and Down's syndrome. Cri-du-chat Syndrome – deletion of genetic chromosome, 5Turner's syndrome.

Congenital infections include HIV, rubella (German measles), chicken pox, syphilis, herpes, toxoplasmosis, Cytomegalovirus; the most common congenital infection leading to hearing loss.

A congenital metabolic disease is also referred to as an in born error of metabolism. Most of these defects are single gene defects, usually heritable. Many affect structure of body parts but some simply affect the function e.g. Phenyl ketonuria, galactosemia.

Endocrine disorders are congenital e.g. congenital hypothyroidism congenital adrenal hyperplasia. Other disorders are muscular dystrophy, cystic fibrosis.

### **Causes of Congenital disorder**

- 1) Use of Antibiotics around the time of conception particularly Sulphonamides, Nitrofurantoin are associated with major birth defects.
- 2) Smoking, Alcohol consumption, Drugs addiction can lead to Foetal Alcohol syndrome, which describes birth defects and other associated problems in infants.

- 3) Risk factors for Congenital Heart diseases are Maternal irradiation, Maternal diabetes, Advanced maternal age, Maternal alcoholism.
- 4) Family history of Chromosomal abnormalities.
- 5) Infectious diseases during pregnancy can lead to defects.
  - a) Rubella can cause defects in brain, heart, eyes and ears of baby and increase the risk of still births.
  - b) Chicken pox can lead to defects in brain eyes skin limbs of baby.
  - c) Measles can increase risk of premature or stillbirths.
  - d) Hepatitis B can cause Hepatitis to the child or it may become carrier of the disease.
  - e) Whooping cough can cause Seizures, Encephalopathy and Stillbirth.

### **Clinical manifestation of some congenital diseases**

- 1) Clinical manifestation of Congenital Heart Disease is Murmur. Murmurs are noted on the first day of birth. A reduced femoral pulse suggests Coarctation. Basal crepitation and peripheral oedema are most reliable signs of congenital heart failure in neonate.
- 2) Feeding difficulties are observed.
- 3) Tachypnoea, Hepatomegaly, Tachycardia are clues for Congenital Heart disease
- 4) Imperforate anus can present as abdominal distension, absence of first meconium stool. Definitive diagnosis can be done by radiographic studies.

### **Screening of Congenital Diseases can be done inutero**

- 1) FIRST Trimester of pregnancy:
  - A) Anomaly scans for Trisomy 21 (Down's syndrome) and Neural tube Defects.
  - B) Maternal serum screening for a) Free beta hcg
  - C) Pregnancy associated plasma protein A (PAPP-A)
- 2) Second Trimester of pregnancy:
  - A) Maternal serum screening –Triple screen = AFP (Alpha feto protein), hcg. Estriol. AFP is typically reduced in Down's syndrome in early II trimester, whereas it is high in Neural tube defects. Maternal serum hcg is twice as high in Down's syndrome. Unconjugated estriol is 25% lower with Down's syndrome.
  - B) Quadruple screen: Triple screen + Inhibin A may increase sensitivity for Down's syndrome.
  - C) Amniocentesis is done for --Detecting chromosomal abnormality.

D) Foetal echocardiography is suggested in patients having family history of CHD, Sonographic anomalies or markers including extra cardiac anomalies, maternal diabetes and infection suspicious scan on screening chromosome aberrations.

E) Additional imaging techniques: Magnetic Resonance Imaging may help specific anomalies; such as agenesis of Corpus Callosum; Posterior fossa cysts; Cerebral cleft.

### Treatment Modalities

Treatment for congenital diseases is based on the type of disorder.

- 1) For skeletal disorders like flat foot etc surgical procedures, artificial limb transplant.
- 2) Imperforate anus: For high defect abdominoperineal resection is done.
- 3) Vaccines can protect against many infectious diseases such as measles, rubella, whooping cough, mumps, diphtheria, tetanus, hepatitis B. Infectious Congenital diseases can be prevented by immunization of pregnant women.
- 4) For cleft palate, surgery is performed which include many specialist such as plastic surgeon, paediatrician, orthodontist, otolaryngologists.
- 5) For CHD surgical correction is done i.e. Blalock Tausig procedure is performed.
- 6) Surgical intervention can correct only few congenital defects but others are not reverted, hence there is need to avoid such births by screening of such diseases.

### Ayurvedic Perspective of congenital diseases

Congenital disorders in Ayurveda can be counted under Aadhyahmik Vyadhi. The disease termed Adibala pravrtta is ascribed to any inherent defect in the semen or the ovum of one's parent, which forms one of the original and primary factors of 'being' and includes leprosy (Kustha), haemorrhoids, Pthisis etc.

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This type is subdivided into two types

- 1) Generated by deranged paternal
- 2) Deranged maternal factor at the time of incubation.

Susruta has explained the congenital or Janma-bala pravratna type usually follows such causes as an improper conduct on the part of the mother during the period of gestation and

embraces such defects or maladies as blindness, deafness, dumbness, nasal voice and such monstrous aberrations of nature as congenital Cretinism and the births of dwarfs and pigmies.

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The Janma-Bala pravrtta can be classified in two types

- 1) Due to action of deranged lymph –chyle (rasakritta) or
- 2) Due to an ungratified desire of mother during gestation or her gratification of any improper longing or conduct during pregnancy (Dauhrdya).

Carak has stated six causes for congenital defects, they are

- 1) Defects in seeds i.e. sperms and ovum,
- 2) Actions associated with the soul,
- 3) Defects in uterus, time of inception
- 4) Imbalance in mental status of mother and father
- 6) Mistakes in food and regime of mother.

Because of these six causes the doshas get viatiated which results in impairment of shape, colour and sensory as well as motor organs of the offspring (Ch. Sh 2/20).

Carak has stated the following factors which cause impairment of pregnancy. If the mother shows addiction to sweet things causes prameha, atisthaulya, addiction to intake of sour things causes rakta pitta and diseases of skin and eyes in offspring, addiction to bitter things makes offspring cachectic, weak and emaciated. Addiction to intake of astringent things makes the offspring gray in complexion, constipated and udavartin (tympanitic). Addiction to wine makes offspring constantly thirsty, short memory and fickle minded (Alpasmriti) (Ch Sh 8/21).

Ayurved also describes sex abnormality, Carak has described eight types of sexual abnormalities 1) Dviretas, 2) Pavenendriya tva, 3) Samskaravaha , 4) & 5) Nar Nari Phandhatva, 6) Vakri , 7) Irsyabhirati, 8) Vatikphanda

These sexual abnormalities are caused by the effects of misdeeds in previous life of the individual.

If pregnant woman resorts to regimes described as the causative factors for the various diseases, her offspring also predominantly suffers from the disease caused by such etiological factors.

### **Treatment for congenital disease in Ayurveda**

Congenital diseases in Ayurveda are considered to be Incurable. But still some congenital diseases such as cleft palate surgeries are described (Ash. ut. 3/25). All the diseases which are Kulodbhava (Familial / Hereditary) are described to be incurable as in case of Sahaja Arsha (Su Ni 7/7).

### **CONCLUSION**

Thus to get desirous offspring with excellent qualities both mother and father should refrain from unwholesome diet and regimes, and they should follow beneficial diet and regime (Ch Sh 8/21).

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In Today's era congenital disorders can be prevented by pre-screening in utero and by taking preventive measures such as vaccination for specific disorders. Congenital diseases can be corrected surgically. But still some chromosomal impairment cannot be cured completely; hence there is a need for taking precautions during gestational period.

### **REFERENCE**

1. Dr.Ganga Sahay Pandey Edited Carak Samita. elaborated by Carak and Drhabala edited Charak Chandrika by Brahmanad Tripathi, Chaukhamba surbharati Prakashan Vol 2 (C.Sh.8)
2. Kaviraj Kunjalal Bhishagratna edited Susruta Samhita by Dr. Laximdhara Dwivedi, Chaukhamba Sanskrit Series Offices Varanasi, Vol 1 (Shu Su 24/5).
3. Susruta Samhita, Dalhan Critic edited by P V SHARMA Chaukhamba Visvabharati Varanasi. Vol II
4. P Astanga Hridayam,Vagabhatta; translated by Prof K.R Srikantha Murthy, Krishna das Ayurved Series 27 Chaukhamba Krishnadas Academy; Varanasi. Vol III
5. Carak Samhita, Cakra pani Dattas Ayurved Dipika, edited by R.K Sharma, Bhagwan Dash, Chaukhmba Sanskrit Series office Varanasi

6. Screening for congenital diseases
7. Prenatal diagnosis of congenital anomalies. Author- T. Todros, E. Capuzzol, P. Gaglioti.
8. Net search- searching for congenital abnormalities.
9. Better Health Channel. Home > Relationship & family > Pregnancy & birth- Pregnancy advice > Immunization & pregnancy.