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Study of congenital anomalies at the tertiary care hospital

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Abstract

Birth of a baby is the happiest moment in the family. Congenital malformations can cause more misery in the family. Genetic anomalies are due to exposure to teratogens in the environment, unknowingly taking teratogenic drugs, nutritional deficiencies such as folic acid, X-ray exposure above 10 Rads during first trimester of pregnancy, maternal infections with TORCH pathogens and syphilis, having personal habits like smoking and chewing tobacco and alcohol abuse. 37 congenital anomalies reported in our study at Special newborn care unit at Chittoor, district head quarters hospital are as follows club foot 10 cases, cleft lip and cleft palate in 7 cases, only cleft lip in 5 cases, meningo myelocele in 3 cases, anotia in 2 cases. Other anomalies reported are duodenal and jejuna l atresia 1, ambiguous genitalia 1, pulmonary hypoplasia 1, skeletal dysplasia 1, dandy walker malformation 1, ectopia vesicae 1, congenital spinal defect 1, Erbs palsy 1, left ear malformation 1 and congenital dislocation of hip bone in one case. Hydrocephalus 3 cases, Meningomyelocele 4 cases, anencephaly 5 cases are other the major anomalies we observed in still born and intrauterine deaths.

Keywords: Hydrocephalus, anencephaly, club foot, cleft lip, cleft palate, congenital anomaly and teratogens

Introduction

Review of literature; Congenital anomaly is a structural, functional or metabolic abnormality that is present from the birth. It can be fatal or result in mental or physical disability. It contributes to significant fetal mortality and infant morbidity and mortality. It can be serious or minor. There are more than 4000 congenital anomalies existing. Some can be cured and some cannot. Incidence is 2-3% in most of them healthy and term babies [1]. Prevalence -3% major congenital anomalies in live birth at birth are reported. And additional 3% diagnosed at later age. 10% minor congenital anomalies in live births [3]. Malformations can be 1. Lumping vs. splitting of outcome, 2. Single vs. multiple defect, 3. Embryonic layer like endoderm, mesoderm or ectodermal defects 4 Syndromes like Downs where one etiology can cause multiple defects in CVS and CNS and finally 5 .Organ system defects. Congenital anomalies account for 15% of perinatal deaths [3]. Central nervous system malformations seen in 39.2%, musculoskeletal system accounted in 14.5% cases and multi system involvement in 18% cases. Incidence of malformation is more in above 35 years pregnant mothers or in multigravida more than fourth gravida [3]. Incidence of cardio vascular defects and gastro intestinal defects were higher in children born to pregnant diabetic mothers [4]. Down syndrome was the commonest multiple malformations. Most of them associated with parental consanguinity [5] Congenital include gross and microscopic malformations, mental retardation, molecular and cellular abnormalities and inborn errors of metabolism more than 80% malformed conspectuses are lost during the embryonic period itself. More than 90% infants die before birth. Congenital malformations exceed intra partum problems and infection problems, as better neonatal care improved the survival of low birth weight babies. Major causes for congenital anomalies are genetic and environmental. In some nutritional deficiency like folic acid, teratogenic effect of medication, psychological and smoking effects [1].

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Anomalies are divided into five broad groups

1. Single gene defects [mutant].
2. Abnormalities of chromosomes.
3. Multi factorial that is interaction between presumed environmental factors and genetic predisposition.
4. Teratogenic factors
5. Idiopathic. [Despite of advance in genetic studies in more than 50% cases cause is unknown]. Mutant genes, chromosomal abnormalities and teratogens identified in 7-8% cases. 25% of anomalies fall into multifactorial category.

Broad division of malformation falls into 4 categories.

1. Pregenesis- during gamete genesis or gonadogenesis
2. Blastogenesis [first 4 embryonic weeks],
3. Organogenesis [fifth to eighth embryonic weeks] finally
4. Phenogenesis [roughly fetal period]

Causes of Congenital Anomalies mostly unknown etiology

1. Polygenic particularly anencephaly, spina bifida, cleft lip and cleft palate, congenital heart disease and pyloric stenosis or sporadic syndrome complexes like CHARGE syndrome, Vater syndrome, Pierre robin syndrome or prune belly syndrome 1. genetic-chromosomal anomalies which can be 1. Numerical like aneuploidy or polyploidy or 2. Structural-like translocation, deletion, duplication, inversion of chromosomes or due to isochromosomes.
2. Environmental-Drugs like Thalidomid –deformed legs and arms, heart, ear, genital and kidney damage [used in early 60s for morning sickness].
3. Alcohol –Fetal alcohol syndrome, poor attention, memory language function, mental retardation, motor in coordination, facial abnormalities like widely spaced eyes and thin lips.
4. Tobacco-low birth weight, sudden infant death syndrome and attention deficit and hyperactivity syndrome... 4. aminopterin-anencephaly, hydrocephaly, cleft lip and palate. 5. phenytoin-facial defects and mental retardation.
5. valproic acid-neural tube defects .cranio, facial, limb and heart anomalies.
6. Trimethadione–cleft palate, heart defects.
7. Lithium in early pregnancy can cause heart malformation.
8. Amphetamines can cause heart defects and cleft lip and palate 8. Tetracyclines can cause teeth discoloration and long bone growth defects.
9. Warfarin –microcephaly and chondroplasia. 10-ACE inhibitors, 11. cocaine and 12 isotretinoin, 13 industrial solvents, 14. organic mercury and lead and others 15. cytotoxic drugs and d-penicillamine 16 poly chlorinated biphenyls and 17. herbicides etc.
10. 3. Maternal infections like – Rubella can cause cataracts, glaucoma, heart defects, tooth abnormalities, and deafness. 2. Cytomegalo virus can cause blindness, microcephaly, fetal death and mental retardation.
11. Herpes simplex –retinal dysplasia, microcephaly and microphthalmia. 4, Varicella –mental retardation and limb hypoplasia.
12. Human immune deficiency virus can cause -growth retardation and microcephaly.

13. Toxoplasmosis-cerebral calcification, hydro cephal and microphthalmia.
14. Syphilis-deafness and mental retardation.
15. Hormones that can be 1 Androgenic –masculinisation of female genitalia 2. Diethyl stilbestrol 3. Maternal diabetes neural tube defects and heart deformities.
16. Physical agents like X-rays-cleft palate, microcephaly, spina bifida and limb effects.
17. Isotopes
18. Hyperthermia

Maternal causes: Obesity can cause heart defects and omphalocele. Other causes include maternal age less than 17 or more than 35, order of pregnancy, geographical location, social class [anencephaly], maternal diabetes, pre diabetes, season of birth, immune related or blood incompatibility like Rh incompatibility. Regional factors like deformation of uterus, oligo hydramnios, ectopic pregnancy, abnormal uterine position, twinning, amniotic bands, and abnormal uterine position of fetus. Maternal deficiency of nutrients like iron, calcium, vitamin B., vitamin C and vitamin D and trace elements deficiency like iodine [teratology factor responsible for cong anomalies et (6). MCA multiple congenital anomalies may be 1. two or more major malformations [neural tube defects, cardiac defect or missing limb] or 2. 3 or more minor malformations [such as abnormal pinna, club foot or syndactyly] cause not known in 40-60% cases, in 7.5% it was single gene mutations, in 6% it was chromosomal anomalies, in 5 % it was attributed to maternal illness or substance abuse [7]. Birth defect is caused when genetic material is damaged or genes misread during organ formation. Sometimes passed in the families or due to teratogens. Causes can be illness in the mother, infections in mother such as rubella, cytomegalovirus, toxoplasmosis, sometimes environmental chemicals or alcohol or medicines consumed by the mother or it can be due to nutritional deficiency like B-complex [8]. Etiology- Normal human development depends on correct chromosome complementation. Usually 22 homologous pairs of autosomes and one pair of sex chromosome, one member of each pair is inherited from each parent. Malformation of chromosome may be due to excess or deficiency of chromosomal material including unbalanced arrangements. Chromosomal material excess or deficit can arise either due to changes chromosomal number or structure [9, 6].

Variations can be 4 types

1. Malformations: abnormal intrinsic development, i.e. Organogenesis [from 3rd to 8 the week of gestation].
2. Disruptions–due to destruction morphological alteration of already formed structures -bowel atresia due to vascular lesions.
3. Deformations- molding of part of fetus for longer period – club foot due to Oligohydramnios.
4. Dysplasia-tissue formation malformations–ex, congenital ectodermal dysplasia [6]. Risk factors include advanced maternal age 35 years and odd, previous gestational history of chromosomal abnormality, history of neural tube defects, and chromosomal abnormality in either parent or a mother with x-linked disease carriage. In these group following investigations required -1. Ultra sound which detect growth parameters 2. Maternal serum screening for alpha fetoprotein which

can detect abnormal neural tube defects.3. Amniocentesis – where needle inserted to amniotic cavity and fluid collected that is biochemically analyzed and subjected to genetic and culture studies.4. Chorionic villous sampling where tissue is aspirated and tested. Problems of measurement of birth defects -- 1. Many congenital abnormalities result in spontaneous abortions and stillbirths 2. Many in live births represent selective survival 3. Many manifest in later stages of childhood eg cardiovascular disease [2]. Strong association between congenital malformations and prematurity noted. National estimates of congenital malformation are important for determining the service planning, monitoring trends and assessing the disease burden [10]. Childhood cancers may be due to intrauterine exposure and due to fetal origin-ex childhood leukemia, neuroblastoma, and brain cancer [2].

Methodology

The Apollo medical college and hospital at Chittoor is conducting in and outpatient services and caesarian operations throughout the year. Conducting roughly 5000 deliveries in a year. During regular antenatal checkups tifa scan is done at 16-18 weeks. Some rural folk admitted as unbooked cases and delivered babies with congenital anomalies. In a retrospective study from January 2016 –to December 2017, these 37 cases were admitted for further treatment at special newborn care unit SNCU .Babies with major congenital anomalies 1. Hydrocephalus3, anencephaly5, we observed were still born or ended in intra uterine death. 37 cases admitted in SNCU were admitted, treated and advised follow ups regularly. Minor anomalies like club foot in 10 cases, cleft lip in 5, cleft lip and palate in 7,meningomyelo cele in3 Anotia in 2, skeletal dysplasia and pulmonary hypoplasia 1, ambiguous genitalia 1, duodenal and jejuna atresia, ectopia vesicae 1, Erbs palsy 1,cong spinal defects 1 and dandy walker syndrome 1, left ear malformation in 1,skeletal abnormality in 1,and congenital dislocation of hip in one case.,

Discussion

60 % of congenital malformations are idiopathic, 2 leaking membranes and continuation of pregnancy, 3 viral infections, 4 congenital syphilis, 5 exposure to drugs or folic acid deficiency .In our study multiple factors involved in congenital malformations. Club foot is corrected by applying casts up to 3 months and physiotherapy. Meningo myelocele, duodenal atresia, ectopia vesica cases were referred to the pediatric surgery department. Erbs palsy is treated with physio therapy, cortisone and B complex. Cleft lip and cleft palate are distinct entities although they are related embryo logically, functionally and genetically. Cleft lip results from failure of fusion of median nasal process and maxillary process. Cleft palate results from failure of palatal shelves fusion. Incidence of cleft lip is 1 in750.incidence of cleft palate is 1.2500 and more common in males. Higher in Asians. Maternal drugs, genetic and syndrome malformation complex attributed. Cleft palate is complicated by feeding problems and needs multidisciplinary approach. Closure of cleft lip done at 3 months through Z plasty operation. Cleft palate is repaired by 1 year as its delay in repair interferes with normal speech development. Recurrent otitis media and hearing loss are the common complication of cleft palate. Feeding is assisted by prosthetic obturator.

Club foot can be congenital in 75% cases, existing as isolated entity. It can be teratogenic due to neuro muscular disorder or positional in other cases. caused by multifactorial inheritance. Calf atrophy, rigid foot and mild hypoplasia of tibia, fibula and foot bones associated. Common in males. Bilateral in 50% cases. Incidence is 1 in 1000.3% more incidences in subsequent siblings, upto 30 % more in affected offspring are reported... Treatment is conservative. Through application of malleable splint and serial plaster casts, which are especially useful in preterm. First position is manipulated, before cast is applied and it is changed at 1-2weeks intervals. By 3months of age clinical and radiological correction occurs. Then casting continued for 3-6 months. In case of failure, operation is required usually before 6-12 months, satisfactory results seen in 80-90% cases.

Conclusion

Genetic examination and screening for Down syndrome, folic acid supplementation, tl-fa scan to exclude congenital anomalies at 18-20 weeks can avoid many congenital defect births; we can better visualize congenital malformation at 24-28 weeks .MTP act 1971 allows termination of pregnancy up to 20 weeks only. Screening for VDRL at 1st semester and 3rd semester can prevent 100% of sequelae of congenital syphilis. Anencephaly can be identified at 14 weeks and we can terminate the pregnancy. Hydrocephalus can cause obstetrical obstruction and was delivered by elective section, baby can survive stent placement .Low anal imperforation can be treated with cruciate incision in anal membranes and regular dilatation by their parents. High anal membrane defect can be treated with 2 stage operation first by colostomy followed by reconstruction. Other surgical cases referred to pediatric surgery department, Vellore. When delivering the after coming head birth injuries like Erbs palsy, fracture humerus, Atlanta axial dislocation or mandibular dislocation are. Commonly observed, this can be prevented by elective ELCS .and trained staff in such circumstances in hospital is necessary.

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