CONGENITAL DISEASES IN THE NEWBORN INFANTS

BY

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Congenital diseases constitute a significant neonatal problem today, particularly in developed countries where the standard of social and health services including obstetric and paediatric and paediatric care, is high (Table I).

TABLE I

<table>
<thead>
<tr>
<th>Birmingham</th>
<th>Sweden</th>
<th>Japan</th>
</tr>
</thead>
<tbody>
<tr>
<td>Total Births</td>
<td>56,760</td>
<td>44,109</td>
</tr>
<tr>
<td>Incidence of major malformations (per 1000 births)</td>
<td>17.3</td>
<td>11.2</td>
</tr>
</tbody>
</table>

*Table I.* Incidence of Malformations per 1000 total births estimated soon after birth (Mckeown & Record, 1960).

While advances in the prevention of many causes of perinatal mortality have been attained, the incidence of congenital malformations, its foetal deaths, neonatal morbidity and mortality, have remained unchanged. Among the causes of Neonatal deaths congenital anomalies rank third in the list.

TABLE II

<table>
<thead>
<tr>
<th>Cause</th>
<th>1951</th>
<th>1961</th>
</tr>
</thead>
<tbody>
<tr>
<td>Immaturity and other prenatal and Natal cases</td>
<td>66.2</td>
<td>48.9</td>
</tr>
<tr>
<td>asphyxia and atelectasis</td>
<td>40.4</td>
<td>44.6</td>
</tr>
<tr>
<td>Congenital Malformations</td>
<td>39.2</td>
<td>36.3</td>
</tr>
<tr>
<td>Birth Injuries</td>
<td>31.5</td>
<td>23.2</td>
</tr>
</tbody>
</table>

*Table II.* Infant mortality, U.S.A. 1951 and 1961 under one year per 10,000 live births. (Department of Health Education and Welfare) U.S.A.
Many congenital malformations are incompatible with extrauterine life and consequently their incidence in still birth is high. Among 5739 products of conception weighing over 500 Gm., McInotsh and co-workers (1954) found that congenital malformations constitute an important cause of still births and neonatal deaths (Table III).

**TABLE III**

<table>
<thead>
<tr>
<th>Incidence</th>
<th></th>
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<tbody>
<tr>
<td>Over-all incidence of malformations</td>
<td>7.5%</td>
</tr>
<tr>
<td>Incidence among antepartum deaths</td>
<td>13.6%</td>
</tr>
<tr>
<td>Incidence among intrapartum deaths</td>
<td>23.3%</td>
</tr>
<tr>
<td>Incidence among Neonatal deaths</td>
<td>29.6%</td>
</tr>
</tbody>
</table>

Table III Incidence of congenital malformations (McInotsh, 1954).

There are many types of congenital diseases which are now compatible with life owing to the recent progress in surgery, anaesthesia, biochemistry and antibiotics.

In the Sudan, although infectious diseases and malnutrition still remain the most major Paediatric problems, congenital diseases are gaining significance owing to increasing awareness of the problem, its methods of prevention and treatment. Accurate vital statistics about the incidence of congenital malformations cannot yet be obtained, but our clinical impression, judging from the large number in our records, indicates that they are uncommon. Some rare congenital diseases such as Morquio's disease have a strikingly high incidence in the Sudan. The common practice of consanguinity is of particular genetic significance as an aetiological factor.

This paper will discuss the most common congenital diseases encountered in Sudanese newborn infants.

**Classification:**

1. **Congenital skeletal diseases:**
   1. Osteogenesis Imperfecta.
   2. Congenital malformations of the Extremities:
      (a) Phocomelia.
      (b) Syndactyly.
      (c) Polydactyly.
      (d) Macrodacltyly.
   3. Achondroplasia.
   4. Gargoylism.
5. Sacro-coccygeal agenesis.
6. Craniosynostosis.
   (a) Craniofacial dysostosis (Crouzon’s disease)
   (b) Scaphocephaly.

(2) **Congenital diseases of the muscles**
1. Arthrogypsis multiple.
2. Congenital absence of abdominal muscles.

(3) **Inborn Errors of metabolism**
1. Phenylketonuria.
2. Galactocaemia.

(4) **Congenital diseases of the Nervous System**
1. Rubella syndrome.
3. Congenital hydrocephalus.
5. Spina Bifida.

(5) **Congenital Endocrine Diseases**
- Cretinism.

(6) **Congenital Heart Diseases:**
1. Transposition of the vessels.
2. Coarctation of the aorta.
3. Aortic atresia.
4. Pulmonary atresia.
5. Tetralogy of Fallot.
7. Dextrocardia.

(7) **Congenital Blood Diseases**
1. Haemolytic Diseases of the Newborn
2. Glucose-6-Phosphate Dehydrogenase Deficiency.
3. Haemophilia.

(8) **Congenital Diseases of the Lymphatic System**
1. Congenital Lymphoedema (Milroy’s Disease)

(9) **Congenital Disease of the Respiratory System**
1. Choanal atresia.
2. Congenital Laryngeal Stenosis.
3. Diaphragmatic Hermia.

(10) Congenital Diseases of the Digestive System
1. Hare-lip and cleft-palate.
2. Micrognathic.
3. Duodenal atresia.
5. Malrotation of the gut.
6. Hirschsprung’s Disease.
7. Imperforate anus and rectal atresia.

(11) Congenital Diseases of the liver and bile.
1. Glycogen disease.
2. Congenital atresia of the bile ducts.

(12) Congenital Diseases of the Skin.
1. Congenital Icthyosis.
2. Congenital Syphilis.
3. Albinism.

Clinical Manifestation in the Newborn Infant.

(1) Congenital Skeletal Diseases.
1. Osteogenesis Imperfecta

This is the intrauterine type of the disease, which manifest in the newborn by extremely thin cranial bones and generalized multiple fractures. The sclerae have strikingly blue appearance. It is definitely hereditary, but there is considerable variation in the degrees of penetrance. Some families showed a dominant genetic defect.

Most infants die in utero, during delivery or soon after. Subarachnoid haemorrhages seem to be the immediate cause of death.

(2) Congenital Malformations of the Extremeties.

(a) Phocomelia. – i.e. great reduction in size of proximal parts of the limb may be of genetic origin. The thalidomide tragedy resulted in a large number of phocomeliae of the upper limbs in Europe. This tranquillizing drug given during the early weeks of pregnancy, the most dangerous period was between 28 to 42nd. day of onset of pregnancy. Other congenital anomalies included atresia or stenosis of oesophagus and duodenum.

(b) Syndactyly, Polydactyly and Macrodactyly can occur alone or in association with other congenital malformations.
(3) **Achondroplasia**

The disease is hereditary, which may be dominant or recessive. The defect involves ossification of cartilage and leads to shortening of limbs. It can be diagnosed by an antenatal radiological picture. At births the clinical picture of short limbs and the relatively large head is diagnostic. The majority of cases are either still-born or die soon after birth.

(4) **Gargoylism.** (Chondro-Osteo – dystrophy)

This is a familial disease, and if one baby is affected in a family, the recognition of another similarly affected new-born infant becomes possible. The fully developed manifestations of hepatosplenomegaly, mental retardation, kyphosis, dwarfism, corneal cloudiness appear later. The disorder is genetically determined being caused by an autosomal recessive disease. The basic metabolic disturbances is now known to be intracellular shortage of mucopolysaccharide, which can be found in the urine in high concentration from birth.

In a family under my care the first-born infant was affected. The following two pregnancies occurred in United States where the father was working in the Sudan Embassy. Aminocentesis was carried out there at the 4th. month of pregnancy and correct diagnosis of the second affected baby and a third normal new-born was achieved at each occasion.

(5) **Sacra-coccygeal agenesis.**

Congenital absence of the Sacrum has a characteristic physical appearance, which is lack of prominence of the buttocks with absence of a groove between them. X-ray shows absence of sacral and coccygeal bones. The defect is not familial.

(6) **Craniosynostosis.**

Congenital premature closure of cranial sutures leads to deformity of head and may cause damage to the brain and eyes. The defect is not common.

(7) (a) Scapocephaly – the head is large and narrow due to premature closure of sagittal suture.

(b) Cranio facial dysostosis (Crouzon’s disease), is characterized by acrocephaly – (the head anteriorly), exophthalmos, external strabismus, hypoplastic maxilla, a beak-shaped nose, short upper lip and protruding lower lip. It is transmitted as a dominant hereditary trait. The condition is progressive from birth.

(3) **Congenital Diseases of the muscles.**

1. **Arthogryposis multiplex Congenita.** (Amyoplasia Congenita).

   The infant is born with fixation of the large joints in extension or flexion and with considerable aplasia of muscle groups. It is a rare disease with predominance in males.
2. **Congenital absence of Abdominal Muscles.**

The appearance of the abdomen is characteristic, the skin is wrinkled, the abdomen bulges with crying and the peristaltic waves are clearly seen. There are associated congenital anomalies, especially in the genito-urinary tract.

3. **Inborn Errors of Metabolism.**

1. **Phenylketonuria.**

This is extremely important condition owing to the necessity of early diagnosis and early dietary treatment in order to permit normal intellectual development and prevent mental retardation.

It is a genetic disorder of phenylalanine metabolism which is recessively inherited. There is absence of the hepatic enzyme, phenylamine hydroxylase, which converts phenylalanine to tyrosine. In its absence there is accumulation of phenylalanine in the serum, and excretion of phenylalanine phenylpyruvic acid and other abnormal metabolic products in the urine.

The infant looks normal at birth and signs of mental retardation may not be suspected before the age of 4 months. Premonitory signs e.g. vomiting, unusual irritability and offensive order of urine may be noticed earlier.

Guthrie's screening test for detecting phenylketonuria in the first week of life is simple and sensitive but a number of false-positive or false-negative results were reported. However early intensive investigations are essential in the newborn infants of known affected families. The characteristic features of blue eyes, blonde or fair hair and mental retardation are late manifestations.

(4) **Galactosaemia.**

Galactose is an important sugar in the diet of the newborn infant. It is combined with glucose as the disaccharide lactose, which forms about 40 per cent of the caloric intake of the breast-fed infant. galactosaemia is caused by deficiency of the enzyme uridyl transferase blocking the conversion of galactose –1– phosphate to glucose –1– phosphate. The infant may look normal at birth but soon after feeding with milk, the infant becomes listless, vomits and begins to lose weight. Jaundice, hepatomegaly, splenomegaly, cataract and mental retardation are late manifestations. Death may result from infection or hepatic failure.

Early detection of this disorder and withdrawal of milk and substitution of galactose-free diet are life saving.

(5) **Congenital Diseases of the Nervous System.**

1. **Congenital Rubella Syndrome.**

The infection in the mother is transferred to the foetus through the placenta.
The first trimester is the most dangerous period. Congenital rubella is an actively contagious disease, the virus can be isolated from nasopharyngeal washings, stools, blood, urine and C.S.F. of the newborn infant. The infant is a source of infection to non-immune contacts. The most common congenital defects are cataracts, cardiac anomalies, especially patent ductus arteriosus, micro-cephaly, deafness and mental retardation. There may be microphthalmus, syndactyly, hopotonia, talipes equinovarus and retinal lesions. The degree of involvement varies widely at birth; small birth weight is a common finding.

2. Mongolism (Down's Syndrome, Trisomy 21)

The mongol can be recognized at birth, although some manifestations are evident later in life. The newborn mongol is small in weight, 20 per cent are premature infants. The skull is small, short and round (brachycephalic). The eyes are slanting with prominent epicanthic folds. The fingers are short and the fifth may be incurved due to hypoplasia of the middle phalanx. There is generalized hypotonia.

Common associated defects are congenital heart disease e.g. Ventricular septal defect in 20 per cent of cases, and duodenal atresia.

The majority of cases have 47 chromosomes (trisomy 21). A few cases have 46 Chromosomes and carry a chromosomal translocation (15/21) which is inherited from one parent, or sporadic.

Recurrence risks are determined by chromosomal analysis.

3) Congenital Hydrocephalus.

Infants who developed severe hydrocephalus in utero may only be delivered with craniotomy. It is easy to diagnose an advanced hydrocephalus at birth or in early infancy but early cases may be missed. Parents may not be aware of a marked enlargement of the head. Surgery (Ventriculo-auricular anastomosis is successful in a large proportion of cases).

4) Congenital amyotonia. (spinal muscular atrophy, Werdnig-Hoffmann Disease).

The infant is hypotonic since birth. Foetal movements are feeble or absent. The infant lies in a froglike position. The muscles are soft and limb movements are feeble. The condition is familial, probably recessive and sexes are equally affected.

5) Congenital Endocrine Diseases

Cretinism is rarely recognized at birth and the cretin may have a large weight at birth and the physiologic icterus may be prolonged. The characteristic manifestations appear later after the second or third month.
6) Congenital Heart Disease.

There is difficulty in the diagnosis of Congenital heart disease in the newborn. During the period of transition from the foetal to post-natal circulation, physical signs such as right ventricular overactivity, second heart sound, colour are governed by the amount of placental transfusion, the rate of constriction of ductus arteriosus and the level of pulmonary artery pressure.

The most important signs are:-
1. Heart murmur—this may not be heard in early infancy.
2. Cyanosis—Other causes of cyanosis in the newborn should be excluded.
3. Congestive heart failure.
4. Tachypnoea, respiratory rate in excess of 50 per minute in indicative Cardiopulmonary disorder.
5. A chest X-Ray is not always reliable.
6. Abnormal electrocardiogram.

The most important Disorders producing frank cyanosis in the newborn period are:

1. **Transposition of great arteries**.

It is characterised by cyanosis from birth, absent E.C.G., and a characteristic egg-shaped heart, which is apparent in X-Ray after the first week of life.

2. **Hypoplastic Right Ventricular Syndrome**:

A small-chambered right ventricle associated with pulmonary valve atresia.

3) **Tricuspid Atresia**:

Gross cyanosis, severe pulmonary atresia, left axis deviation and left ventricular hypertrophy.

4) **Tetralogy of Fallot**.

This is the most important malformation which causes cyanosis late in the newborn period.

The initial signs may be cyanosis or dyopresea or both on crying, blue spells, a loud ejection systolic murmur along the left sternal border. E.C.G. shows abnormal right ventricular hypertrophy. X-Ray chest may be normal or may show right aortic arch.

Cardiac Malformations producing congestive Heart Failure early in the Newborn Period (Rowe, 1970).

1. Hypoplastic left heart syndrome. The most common form is aortic atresia with or without mitral atresia. Congestive heart failure usually appears in the 2nd. day of life and the average age at death is 4 and 5 days.
2. Contracted form of endocardial fibro-elastosis. The aortic valve may be
stenotic, but the aorta is normal. In both forms the heart is very large on X-Ray.
3. Coarctation of Aorta. The pulses in the legs are frequently absent, or faint.
There may be other associated malformations.

5) Congenital Blood Diseases:

1. *Haemophilia* can present in the neonatal period as deep bleeding (giant
cephalhaematoma) or as prolonged superficial bleeding from puncture wounds
or circumcision.
2. *Factor XIII Deficiency*. May cause bleeding from or around the umbilical
cord.

6. Congenital malformations of the lymphatic system.

Congenital lymphoedema (Milroy’s disease) Oedema of one or more of the
extremities are often seen at birth. There is no pain or tenderness. The disorder
is due to congenital abnormality of the lymph channels. It is transmitted as
recessive trait in most cases. Rarely lymphoedema is associated with chylous
ascites.


1. *Choanal atresia*. If both nasal passages are blocked by a membranous,
cartilaginous or bony septum, severe respiratory distress occur since birth which
need immediate intervention to establish an airway.
2. *Congenital Laryngeal stridor*. Is relatively common in the neonatal period
and it usually disappears by one year of age.
3. *Congenital Diaphragmatic Hernia*. In some cases, herniation of abdominal
contents into the thoracic cavity causes severe respiratory embarrassment in
the newborn infant and it constitutes medical and surgical emergency. In
other cases the hernia is detected later in infancy.

8) Congenital Diseases of the Digestive System. *Intestinal Atresia*. 50% occur
in the ileum and 25% in the duodenum. There is increased frequency of
duodenal atresia in Down’s syndrome, and in association with maternal hyd-
ramnios. Treatment is surgical.


1. Albinism.
   Generalized albinism is an inborn defect in the formation of the pigment
melanin. Owing to the absence of activity of the enzyme tyrosinase melanocytes
cannot form melanin. It is transmitted as an autosomal recessive.
2. *Harlequin Foetus* is an extreme form of congenital Ichthyosis, who usu-
ally die soon after birth.
Congenital Tremors

Sacrococcygeal Teratoma. This is a tumour that appears at birth. It is firm and extend downwards and externally. Treatment is immediate excision.

Teratomas are believed to arise from secluded embryonal cells which have primitive potentiality for excessive growth.

3. Congenital Syphilis. It is now extremely rare in most cities in the Sudan where ante-natal care is carried out. The only cases of Congenital Syphilis I saw were in Port Sudan in 1955. The most characteristic manifestation in those cases was pseudoparalysis and X-Ray of bones showed osteochondritis and periositis.

Prevention of Congenital Malformations:

The chance that any pregnancy will result in a serious malformation or mental retardation is about 2 or 3 per cent, Hecht and Loverien (1970).

The majority of congenital malformations are the result of the genetic and environmental factors. Thus the plans for prevention should include:

1. genetic counselling. This is particularly important in the Sudan where consanguinity is the rule in marriages.
2. Avoidance of infection especially rubella and other viral infections during pregnancy particularly the first trimester.
3. Avoidance of irradiation any time during pregnancy.
4. Drugs, which may have teratogenic effects, should be avoided especially early in pregnancy.

Fig. 1. Osteogenesis imperfecta
REFERENCES

Rowe R.D. (1970) Serious Congenital Heart Disease in the Newborn Infant. Diagnosis and management.