OTHER BIRTH DEFECTS

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Other birth defects

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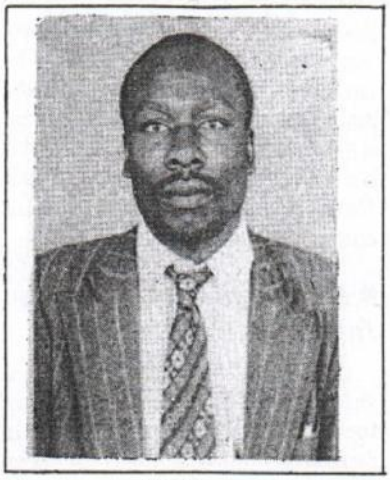
Congenital malformations represent a major cause of infant death and create a most urgent problem of medical and social importance. This is on account of physical disability in the survivors and beeause of primary or secondary mental defects as often associated with such malformations. This creates the need for suitable long term hospital accommodation for such patients, corrective surgery both timely and multidisciplinary, and adequate physical and psychotherapy. It is thus important that awareness be enhanced and deeper insight created into this question of birth defects. In this article, the last on the series of birth defects, I shall discuss the other malformations of systems other than those of the nervous, heart and muscle-skeletal systems.

• *Defects*

These are common and more frequent in males.

Cleft Lip

This malformation can be unilateral or bilateral and the nose may be affected. If the parents are healthy but one child has a cleft lip, chances of a second child having the defect is 4-7 in every 100 children. If one of the parents is affected, chances



Dr. Ogeng'o: "Defects which children are born with represent a major cause of infant death and create a most urgent problem of medical and social importance."

that a second child will be affected is between 11 and 14 per 100 children.

Cleft Palate

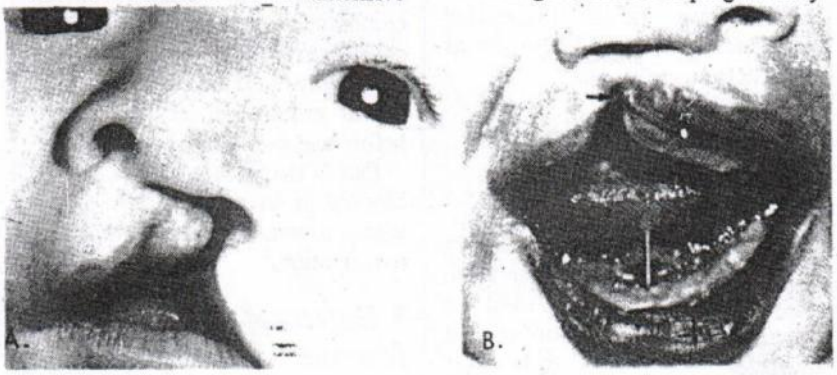
This occurs in 2 — 3 per 100 children and is associated with extensive irregular teeth arrangement. These defects interfere with sucking, proper feeding and later development of speech. Infections of the nasopharynx and ear are commoner in the victims. Both of these conditions are fortunately amenable to successful surgical correction once the timing is right.

• *Defects of The Eyes*

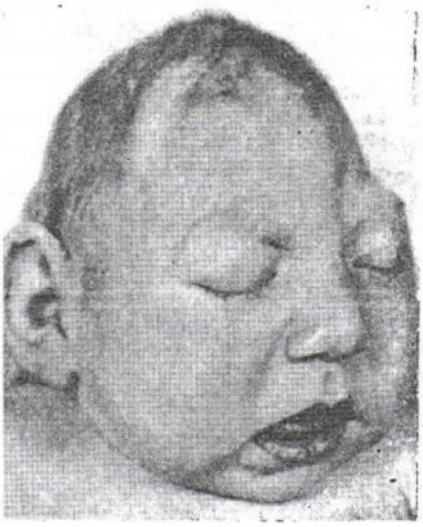
The complex structure of the eye and thus the complicated process of its development increases its sensitivity to harmful external agents. Viral infections, syphilis, vitamin deficiencies and excesses, and x-rays, all may be responsible for the numerous eye abnormalities. These abnormalities can affect the eyeball, part of it, or the surrounding structures. Some examples are illustrated by the pictures below.

• *Defects of the Ears*

Damage to the developing ear may



Photographs illustrating clefts of the lip.



Photograph showing the cranium bifidum - one of the malformations of the face and skull.

be caused by drugs such as streptomycin, commonly used in the treatment of Tuberculosis and taken in pregnancy. Infections such as syphilis and German measles, mumps, influenza and Herpes that may affect the mother in pregnancy may also cause damage to the foetus' developing ear.

These defects may affect the external ear, in which case the entire ear may be absent, it may be too small, too large, or shaped abnormally. Hearing however is not affected.

When the internal ear is affected, the child is most likely going to be deaf and such children are recognised by decreased vocalisation, changes in behaviour, monotonal vocalization and extreme visual attentiveness.

Small discharging 'holes' anterior to the ear called Prehelicine Fistualae occur quite frequently as other defects of the ears.

• *Malformations of the Digestive System*

These are numerous and affect various parts of the alimentary canal. Only the common ones will be mentioned. Most of them can be corrected surgically and early diagnosis can be life-saving. Examples of these defects include:

Congenital Oesophageal Atresia.

This is commonest in this group. In this defect, the lumen of the oesophagus is not open and therefore food cannot pass. The child will thus vomit after any feed. This defect is commonly associated with communication between the oesophagus (gullet) and the trachea (airway) in which case food gets into the lungs causing choking.

Congenital Hypertrophic Pyloric Stenosis

In this defect the muscles at the outlet of the stomach called pylorus are too strong and cause obstruction. Persistent projective vomiting is the first symptom of this defect and begins in the second week of life. The vomiting dehydrates and malnourishes the child. If treatment is not sought immediately, the child is likely to die from dehydration. However, surgical correction is almost always successful.

• *Defects of the Small Intestines*

Congenital umbilical hernia is characterised by the enlargement of the umbilical ring, resulting in protruding mass covered by skin. This occurs very commonly among Africans and in some communities in Kenya, it is regarded as a sign of beauty. In a more serious defect, called Omphalocele, the intestines and parts of the liver, heart or stomach may be found outside of the abdominal cavity covered by a thin transparent membrane. Most victims die before intervention is attempted. In some Kenyan communities, such babies are considered a bad omen and are commonly believed to be due to sexual misconduct of the mother before and or in pregnancy.

Part of the small intestines may be blocked in what is called intestinal atresia presenting with vomiting and constipation.

• *Defects of The Large Intestines*

One of the common defects in this category was named after the person who first described it, "Hirschsprung". This defect is characterised by the absence of the peristaltic waves in part of the large intestines and manifests as constipation and abnormal dilation of part of the gut. A baby suffering from this condition has a huge abdomen and looks grossly wasted.

• *Imperforate Anus*

This is a common defect and it occurs in one in 5,000 live births. The baby is unable to pass stool since he does not have an anal opening. Only the doctor can decide on the extent and severity of the problem. Fortunately in the absence of other defects, it is amenable through successful surgical correction. If your child does not pass stool in the past 45 hours after birth, you may be correct to suspect he has an imperforate anus.

• *Defects of The Liver*

The commonest defects affect the apparatus secreting and transporting bile. The commonest presentation is a progressive yellowing of the eyes, called jaundice. This condition, referred to as bilary atresia is of two main types; occlusion outside and within the liver can be relieved surgically while that inside is extremely difficult. If you notice your baby's eyes turning yellow, consult your doctor.

Defects of The Respiration System

These are varied and range from disorders of the nose, larynx, trachea and the lungs. The severe defects kill soon after birth and the other present with difficulty in breathing. Again, only the doctor by careful examination and investigations can place the defect and therefore children presenting with breathing difficulty soon after birth are safer in the hands of a doctor. Their defect may be due to a correctable cause.

• *Defects of The Urination System*

These range from defects of the kidneys all the way down to the

urethra. The kidneys may be absent or poorly formed or in the wrong place. When the latter is the case, the baby presents with inability to form urine and thus waste products accumulate and derange the body functions. Kidneys in the wrong place may still function normally but cause effects similar to those of a mass pressing on the other vital structures.

In extrophy of the bladder, the abdominal wall and anterior part of the bladder do not develop. The posterior wall of the urinary bladder is exposed to the outside. Surgical repair is possible and commonly successful.

The urethra on the other hand may be narrow in different positions or may be completely closed.

• *Defects of The Male Genital System*

Undescended Testes

This is observed in one out of ten boys. However, spontaneous descent occurs and in adulthood only about 3 in every 1000 still have undescended testes. If a testis remains undescended, the process of manufacturing sperms is impaired and one may become frankly sub-fertile. The undescended testis is also more prone to cancerous change. Congenital absence of the testis has also been known to occur.

Congenital Indirect Inguinal Hernia

This refers to profusion of the intestines into the scrotum due to a defect of development. The hernia occurs more frequently on the right. Surgical repair is almost always successful.

Congenital Hydrocele

This may be due to faulty or delayed development of the lymphatic drainage and presents with an abnormally enlarged testis that appears to contain fluid. The defect is amenable by corrective surgery.

Defects of the Penis and Urethra:

Absence of the penis is rare. Defects of the urethra usually involve its position. The commonest defects are called Hypospadias. In this group, the urethra opens on the under side of the penis at different positions. The urethra may also open on the upper part of the penis. These are called Epispadias. Both Epispadias and Hypospadias can be surgically corrected usually to facilitate copulation.

• *Defects of Female Reproductive System*

Defects of the internal organs are usually difficult to detect until much later in life. They can be suspected when a growing girl shows abnormal growth both in structure and function. Only the doctor having done several specialised investigations can make a diagnosis. These investigations include hormonal studies and specialised x-rays. Any of the organs from the ovary through to the vagina can be affected.

A common defect is an imperforate hymen. This remains undetected until the onset of menstruation when there is accumulation of blood in the vagina. This can be relieved by surgical excision of the hymen. In some women, there may be a small opening to allow menstrual flow but inadequate for successful coital intromission. Again, surgical excision makes sexual intercourse subsequently successful. In extreme cases, the full length vaginal canal may be absent. Corrective surgery becomes more complicated but is nonetheless possible.

The concept of Hermaphroditism has been discussed in another issue of this magazine. In this condition, the individual possesses both female and male organs.

• *Multiple Malformation Syndrome*

This refers to the co-existence of more than two malformations in the same individual. It must be understood that the effects of the teratogens most probably affect more than one system and whereve r one malformation is detected, the possibility of malformations must be considered. In rare cases however, multiple malformation may be associated with tumours. It is possible that the causes of malformation may rarely be associated with tumors. Proof for such co-relation in human pathology is still wanting.

• *Conclusion*

Over the past 30 years, the importance of structural defects and their theoretical and practical significance has been increasingly realized. These structural defects are recognised causes of a substantial proportion of deaths before and or after birth all over the world. Indeed it is estimated that at least 20 per cent of the fertilized human zygotes are lost during early pregnancy and that the incidence doubles by the end of the first year with defects not diagnosed at birth. This series of articles of which this is the last is hoped to have illustrated several points:

- that although the exact causes of these defects are not known, the interaction between the environment and the genetic make-up in the causation is certainly important and that we can each contribute in reducing the incidence of birth defects.
- that although all systems are emenable to birth defects, the more complex systems are at a higher risk.
- that the defects vary from system to system and some show regional and racial bias.
- that rapidly advancing technology is widening the horizons of corrective surgery as much as it renders early diagnosis possible.
- that the non-medical population can assist in early detection of some of the defects and thus contribute significantly to salvage measures.

It is against this background that I wish to end by reiterating that if you know of a baby with any structural deviation, consult your doctor now, for tomorrow might too late.