TITLE: MAJOR CONGENITAL MALFORMATIONS IN DAR ES SALAAM (TANZANIA)

AUTHOR: DR. JOYCE M. MGONE: M.D. (DAR)

SUPERVISOR: DR. A. YOHANI: M.B. Ch.B. (E.A.), D.C.H. (GLASGOW), M.R.C.P. (U.K.),
SENIOR LECTURER IN PAEDIATRICS AND CHILD HEALTH
UNIVERSITY OF DAR ES SALAAM

DISSERTATION

A dissertation submitted in partial fulfilment for the award of the degree of Master of Medicine (Paediatrics) of University of Dar es Salaam.

CERTIFICATE

This is to certify that this dissertation is the result of my sole effort.

DECLARATION

I declare solemnly that the whole or part of this work has not been submitted for a degree in any other university.

TABLE OF CONTENTS

1. SUMMARY

Ninety eight cases of major congenital malformations seen in Dar es Salaam over a period of ten months are reported. The pattern of major congenital malformations appears to be similar to that of other places. Three rare malformations one of which has been reported from other places are described.

Parity alone is not associated with congenital malformations but there is a strong relation between advanced maternal age and trisomic syndromes. Also noted is the high frequency of malformations of the musculo-skeletal system in young mothers of low parity. In the study it was also noted that the aetiology of congenital malformations cannot be identified in majority of cases.

Although there was great variation in the type of congenital malformation, initial maternal reactions were similar. Mothers' knowledge about the causes of congenital malformations was minimal. The majority of mothers whose babies had treatable conditions were anxious to have them treated early and continued to care for them. Birth of an abnormal child does not seem to create fear for subsequent pregnancy.

2. INTRODUCTION

Congenital malformations are structural defects present at birth. They may be single or multiple, gross or microscopic, external or internal, hereditary or non-hereditary, familial or sporadic (Warkany, 1975).

There are major and minor malformations. According to Marden and Co-workers (1964), a major malformation is one which had adverse effects on either function or social acceptability of the individual while minor malformation is one which is neither of medical nor cosmetic consequence to the patient. Minor malformations may be taken as a phenotypic variation but when two or more are present it may be a sign of a major malformation (Marden etal, 1964). They may be internal, external or in combination. Internal malformations may escape recognition at birth since they require the use of other diagonistic aids like endoscopic examination and x-rays whereas external malformations can be seen just on inspection. There are many malformations that do not manifest themselves and are not detectable in the newborn children. McIntosh and colleagues (1954) observed that incidence figures of congenital malformations double when the search is extended beyond the first year of life.

True incidences of congenital malformations are difficult to obtain and therefore only estimates are usually given. No two incidence studies are usually the same, for example series that include minor malformations give a higher incidence. Simpkiss and Lowe (1961) found an incidence of 5.4 percent in Africa newborns in Kampala. Khan (1961) in Nairobi found a rate of 1.79 per cent but he had excluded minor malformations.

Some malformations are not detected at birth and this as well affects incidence studies. McIntosh and Co-workers (1954) found an incidence of major malformations to be 7.5 per cent, their study included evaluation for a period of five years. The racial make up of the study population may also influence the incidence rate of a particular type of malformation. Polydactyly for instance is a minor malformation and is said to be very common amongst the negro race (Altemus and Furguson, 1965).

The incidence of congenital malformations in Dar es Salaam is not known. According to Okeahialam (1974), Dar es Salaam probably has a lower incidence as compared to other parts of the world.

In this study, the pattern of major congenital malformations in African newborns has been studied. A previous study of Okeahialam (1974) on the pattern of congenital malformations in Dar es Salaam included both minor and major malformations.

The cause of a congenital malformation may be hereditary or environmental and sometimes a combination of both (Sucheston, 1973). Abnormal gene or genes may be inherited from one or both parents or may arise sporadically through mutation (Warkany, 1961). Genes of this sort producing congenital malformations may be dominant or recessive. Conditions caused by dominant genes are for example achondroplasia, cleidocranial dysostosis, lobster-claw deformity of hands and feet and osteogenesis imperfecta. Examples of malformations caused by recessive genes include chondrodystrophia calcificans congenita, some forms of infantile polycystic kidneys and osteocondrodystrophy.

Chromosomal aberrations are known to cause congenital malformations. For example in the regular Down's syndrome there is an extra chromosome on chromosome 21. Lejeune and Collegues (1959) demonstrated this extra chromosome in mongolism. Stewart and Co-workers (1969) examined chromosomes of 184 children with congenital malformations ascertained among 2,500 consecutive live births. They found that 4.3 per cent had abnormal karyotypes. In their study they wanted to find out the frequency of chromosomal abnormalities in children with congenital malformations. Sergovich and Collegues (1969) in their study of chromosomal aberrations in 2159 consecutive newborn babies found gross chromosomal abnormalities to be present in 0.48 per cent. These abnormalities generally

consisted of trisomy of whole chromosomes and some were due to structural variation.

In this study, no chromosomal analyses were done because of lack of facilities but some chromosomal disorders were recognized by finding two or more minor malformations since the finding of minor anomalies may aid in the recognition of known patterns of multiple anomally such as one of autosomal trisomy syndromes (Marden et al, 1964).

Rubella infection is probably the best established environmental means by which a syndrome of congenital malformations may be induced. Sheridan (1964) in a final report of a prospective study of children whose mothers had rubella in early pregnancy found that 15 percent had major malformations mainly of the eye, ear and heart. Minor abnormalities were also present in 16 percent of the children. In another study by Sever and co-workers (1964), 10 percent of women with clinical rubella in the first trimester of pregnancy gave birth to children with congenital rubella syndrome which was recognized within the first month of birth. 0.6 percent of the patients with first trimester exposure who were asymptomatic had children with congenital rubella syndrome. Cytomegalovirus is a rare cause of congenital malformation. Malformations which may result from congenital cytomegalovirus infection include microcephally, hydrocephally, microphthalmia and microgyria (Hanshaw, 1966). Hanshaw (1966) also demonstrated cytomegalovirus complement

fixing antibodies in serum of 44 per cent of 41 infants and children with microcephally.

Infection with the protozoa toxoplasma gondii during the second trimester of pregnancy may also cause congenital malformations. Malformations were seen in 20 per cent of cases of congenital toxoplasmosis (Eichenwald, 1957). These malformations included microcephally, microphthalmia and hydrocephalus.

Drugs affect the embryo during the first trimester of pregnancy. The wide spread use of Thalidomide in Germany in the 1960s lead to a great increase in limb malformation. Lenz (1962) reported 52 malformed infants whose mothers had taken thalidomide in early pregnancy. Drugs which definitely cause congenital malformations are Thalidomide (Lenz, 1966), Cyclophosphamide (Greenberg et al 1964), and Nor-ethisterone. Levy et al (1973) found that of 76 mothers of infants with transposition of great vessels, 6 had received hormones for threatened abortions. Anticonvulsants are also associated with a high incidence of cleft lip and palate, congenital heart diseases and digital hypoplasia (Smithells, 1976).

Exposure to ionizing radiation can also cause congenital malformations. The development of congenital anomalies in the foetus following therapeutic x-ray radiation to the gravid

maternal pelvis has been well known for many years. Births of microcephalic, mentally retarded children have resulted from the accidental over exposure of women in the first trimester of pregnancy to x-ray radiation given as treatment for pelvic disease (Murphy, 1947). In a study on anomalies occuring in children who were exposed in utero to the atomic bomb in Hiroshima Plummer (1952) found microcephally with mental retardation was the most frequent anomally. Microcephally, mongolism mental retardation, hydrocephalus, spinal cord anomalies, cleft palate, abnormal limbs , microphthalmia, cataract, deformed ears are some of the major anomalies which have been found in humans exposed in utero to radiation (Brill, 1964). On the other hand it is claimed that no evidence exists which links diagnostic radiography and congenital malformations (Brill and Forgotson. 1964).

The identification of an infective and chemical teratogen may not be easy. In a retrospective study it may be impossible to establish with certainty that a drug was taken or an infection occured. In this study an attempt was made to identify some of these environmental factors causing congenital malformations. A history of medication, x-ray exposure, rubella infection, contraceptive use, smoking and alcohol intake during pregnancy was asked for in every mother. An attempt was also made to identify some of the hereditary causes

Racial differences are also associated with a high incidence of particular types of malformation. The incidence of anencephally is said to be low in Africans (Penrose, 1957). Simpkiss and Lowe (1961), reported no baby with anencephally in 2,068 newborn Africans in Kampala, Uganda. Other factors associated with congenital malformations are birth order and maternal age, (Carter, 1971), seasonal variation (Slater et al, 1964) and social class. These factors were also looked for in this study.

Congenital malformations can be explained on scientific bases but in some societies where many parents have little or no scientific knowledge, there may be many beliefs and superstitions on this subject. Even from history, birth defects have been associated with superstitions (Warkany, 1963). They were attributed to activities of the Gods either as a punishment for man's sin or a warning against disasters. They were also attributed to crossing of species and maternal impressions during pregnancy.

The presence of an abnormal child in a family may bring a lot of stress. The presence of such a child may result in unwanted reactions from various relatives, they may blame each other through false concepts of the effect of heredity or previous disease (Wilson, 1963). It may be important to know

the parents' social attitude on their malformed child because this can help in the management. The parents may have wrong concepts on the cause of the malformation and they may want to know the chances that a second child may be abnormal. In this study therefore, I have also attempted to find out the mothers reactions and attitudes following birth of an abnormal child.

There is scanty literature on congenital malformation in East and Central Africa. In a previous study by Okeahialam (1974) on the pattern of congenital malformations in Dar es Salaam no attempt was made to identify some of the aetiological factors. And also, no social study has been done on this subject in Dar es Salaam. In this study therefore, the author has attempted to determine the pattern and frequency of major congenital malformations in the African newborns, their aetiological factors and has looked into the social aspect of the problem.

3. OBJECTIVES

The objectives of the study were as follows.

3.1: To determine the pattern of gross major congenital malformations in African newborns clinically recognizable at births.

3.2: To identify and describe the rate malformations that have been reported in literature.

3.3: To determine the maternal age and parity of mothers of malformed babies.

3.4: To identify some of the aetiological factors of congenital malformations in the study population.

3.5: To assess the mothers reaction, knowledge and social attitude towards the abnormal baby.

4. MATERIAL AND METHODS

4.1: THE STUDY POPULATION

The study was carried out in the Neonatal unit and in the follow up outpatient clinic within the Muhimbili Medical Centre. Included in the study were all live born malformed babies who were admitted in the unit during the period of October 1981 and July 1982.

This is the only unit acting as a tertiary centre in Dar es Salaam where all neonates with problems noted at birth or thereafter are immediately referred for special care; this includes those with apparent congenital anomalities.

There are five major hospitals with maternity wings in Dar es Salaam, one of which is privately owned by a religious sect. There are also three private maternity homes. The private groups charge a nominal sliding scale fee hence they cater mainly for the well-to-do minority of the population. The two major government hospitals serve mothers without risk factors while the maternity wing in Muhimbili generally admits mothers with abnormal or at risk pregnancies. There are 416 maternity beds in the whole of Dar es Salaam with an average delivery of 35 000 babies in a year. Because of the pressure of beds the mean confinement period is 48 hours. Therefore, only newborn babies with major malformations apparent at birth were included in the study.

17 babies included in the study were born on the way to hospital or at home but were brought to hospital because of the malformation. A few malformed babies were picked up in the unit during the routine physical examination of the newborn on admission.

4.2: METHODS

All babies with congenital malformations who were referred to the neonatal unit from all over Dar es Salaam were examined by the author. The initial screening for congenital malformations was done by the midwife or a doctor who had assisted in the delivery of the baby. A number of babies were admitted with a definite diagnosis but 31 had been referred as abnormal babies without a diagnosis. Therefore a thorough systematic examination was done by the author and any abnormality was recorded on a standard proforma as shown in the appendix.
Radiological investigations were also performed whenever indicated. No autopsies were performed. In one baby the true diagnosis was reached after a laparatomy for a suspected intestinal malformation which had resulted in intestinal obstruction.

All mothers of malformed babies were interviewed by the author using a standard questionnaire while in the ward and further interviews were done in the follow up outpatient clinic.

The period of follow up ranged between 2-9 months.

Controls for the study group were chosen from normal babies in the unit who had been admitted for care and were matched for age and sex. The same questionnaire was used in the control group.

5. RESULTS

During the period of study, there were 4,575 admissions in the Muhimbili neonatal unit, 98 of which had major congenital malformations.

The sex distribution was 49 boys and 49 girls with a male to female ratio of 1:1. 50 of these malformed babies were born at Maternity Block of Muhimbili Medical Centre and the rest were referred from outside but within the city of Dar es Salaam.

74 had single and 24 had multiple malformations.

5.1: THE PATTERN OF MAJOR CONGENITAL MALFORMATIONS

The pattern of major congenital malformations was studied. Malformations were commonest in the musculo skeletal system. The gastro-intestinal malformations were as frequent as the central nervous system malformations. None of the babies had a malformation of the cardiovascular system as the only defect. Table I shows the frequency of major congenital malformations in the different systems.

TABLE I

THE FREQUENCY OF MAJOR CONGENITAL MALFORMATIONS IN DIFFERENT SYSTEMS

SYSTEM	TOTAL NO.	PERCENTAGE
Musculo-skeletal	31	31.6
Central nervous	17	17.3
Gastro-intestinal	16	16.3
Trisomy syndromes	14	14.3
Urino-genital	5	5.1
Other unclassified malformations	15	15.3
T O T A L	98	100

The frequency of major malformations affecting different systems are shown in Tables II - VI.

TABLE II

THE FREQUENCY OF MAJOR MALFORMATIONS OF THE MUSCULO-SKELETAL SYSTEM

TYPE OF MALFORMATION	NO.	%
Talipes equinovarus	15	48.4
Transverse Limb reduction	6	19.4
Polydactyly	3	9.7
Arthrogyroposis multiplex congenita	4	12.9
Lobster claw deformity	1	3.2
Absent radius	1	3.2
Achondroplasia	1	3.2
T O T A L	31	100

TABLE III

THE FREQUENCY OF MAJOR MALFORMATIONS OF THE CENTRAL NERVOUS SYSTEM

TYPE OF MALFORMATION	NO.	%
Spinabifida cystica	12	70.6
Microcephally	3	17.7
Microcephally and occipital encephalocoele	1	5.9
Encephalocoele	1	5.9
T O T A L	17	100

TABLE IV

THE FREQUENCY OF MAJOR MALFORMATIONS OF THE GASTRO-INTESTINAL SYSTEM

TYPE OF MALFORMATION	NO.	%
Harelip and cleft palate (unilateral)	9	56.6
Bilateral harelip and cleft palate	2	12.5
Anorectal malformation	5	31.3
T O T A L	16	100

TABLE V

CONGENITAL SYNDROMES AFFECTING MULTIPLE SYSTEMS

TYPE OF SYNDROME	NO.	%
Down's	6	33.3
Trisomy 18 phenotype	5	27.8
Trisomy 13 phenotype	3	16.7
Beckwith-Wieldeman	3	16.7
Pierre-Robins	1	5.6
T O T A L	18	100

TABLE VI

THE FREQUENCY OF MAJOR MALFORMATIONS OF THE GENITAL URINARY TRACT

TYPE OF MALFORMATION	NO.	%
Hypospadius	3	60.0
Prune Belly Syndrome	2	40.0
T O T A L	5	100

In the musculo-skeletal system, talipes equinovarus was the commonest malformation. Spinabifida cystica was the commonest malformation affecting the central nervous system (Plate 7). Harelip and cleft palate was the commonest external malformation of the gastro-intestinal tract. Down's Syndrome and Trisomy 18 occured with the same frequency in the group of Congenital Syndrome affecting multiple systems.

Table VII shows the miscellaneous malformations.

TABLE VII

MISCELLANEOUS MAJOR CONGENITAL MALFORMATIONS

MALFORMATION	NO.
Cystic hygroma	2
Choanal atresia	1
Facial strawberry nevus	1
Microtia	1
Congenital rubella	1
De Lange syndrome	1
Microphthalmia	1
Other unclassified multiple malformations	3
T O T A L	11

5.2. RARE MALFORMATIONS

Three babies were seen with rare malformations (Plates 17-19). Table VIII shows the different abnormalities in each of these babies.

TABLE VIII

ABNORMALITIES SEEN IN 3 BABIES WITH RARE MALFORMATIONS

CASE NO.	SEX	BIRTH WEIGHT IN KGS	ABNORMALITIES	DIAGNOSIS
36	M	1.970	Imperforate anus, short thighs, 3 digits on right hand, left foot - everted, short 1st toe, absent 5th toe, Genu recarvatum, Small pelvis	Multiple Anomalies
46	F	1.530	Microcephally, Microphthalmia, Collodion Skin, Rocker bottom feet, toes medially deviated, flexion deformity and contractures of the interphalangeal joints.	Trisomy Syndrome and Collodion skin
55	M	2	Microcephally, thick eyebrows meeting at midline, long eye lashes, short hairy forehead, hairy back, micrognathia, fish mouth, short forearm, contracture of the shoulder and elbow joints monodactyly, absent radius, small penis, underdeveloped scrotum, undescended testes, nostrils pointing upwards.	Cornelia de Lange syndrome

5.3. MATERNAL AGE AND PARITY

The mean maternal age in the study group was 23.9 years and in the control group 26.1 years. The age difference between the two groups was found not to be statistically significant. The distribution in parity was also not statistically significant. Table IX shows the frequency of major congenital malformations by maternal age.

TABLE IX

THE FREQUENCY OF MAJOR MALFORMATIONS IN DIFFERENT SYSTEMS BY MATERNAL AGE

MALFORMATIONS	AGE IN YEARS					
	15-19	20-24	25-29	30-34	35-39	40-45	TOTAL
Musculo-skeletal system	6	12	9	2	1	1	31
Central nervous system	3	3	3	6	1	1	17
Gastro-intestinal system	5	2	7	1	0	1	16
Trisomy syndromes	0	2	1	4	4	3	14
Urinogenital system	1	3	1	0	0	0	5
Other unclassified multiple malformations	1	1	3	1	1	1	7
Others	0	5	1	2	0	1	8
TOTAL	16	27	25	16	7	7	98

x^2 p = 0.03

When malformations of the musculo-skeletal system and trisomy syndromes are considered alone,

x^2 p = 0.002 0.01, the results are highly significant.

Table X shows the frequency of the malformations by parity.

TABLE X

THE FREQUENCY OF MAJOR MALFORMATIONS IN DIFFERENT SYSTEMS BY PARITY

MALFORMATIONS	PARITY				
	1	2-4	5-7	8-10	Total
Musculo-skeletal system	6	19	3	3	31
Central Nervous systems	3	7	7	0	17
Gastro-intestinal	7	4	3	2	16
Trisomy syndromes	0	3	7	4	14
Urinogenital system	1	4	0	0	5
Other unclassified multiple malformations	1	3	2	1	7
Others	3	1	3	1	8
TOTAL	21	41	25	11	98

x^2 $p = 0.05$

Malformations of the musculo-skeletal system are far commoner in mothers of low parity whereas Trisomy syndromes increase with maternal parity. However, these results are not statistically significant.

5.4: ANALYSIS OF POSSIBLE AETIOLOGICAL FACTORS

The possible aetiological factors were identified in 25 cases.

5.4.1: CHROMOSOMAL DISORDERS

Since no chromosomal analysis were done, the diagnosis was only clinical according to the atlas by de Grouchy and Turleau (1977). The following chromosomal disorders were recognized.

Down's Syndrome	- 6
Trisomy 18 Phenotype	- 5
Trisomy 13-15 Phenotype	- 3
TOTAL	14

5.4.2: HEREDITARY/FAMILIAL MALFORMATIONS

There was a positive family history of the same malformation in 7 babies.

5.4.2:1 HARELIP AND CLEFT PALATE

Three babies had a positive family history of harelip and cleft palate. Of these three, one had bilateral harelip and cleft palate (Plate 9) and had a similarly affected elder brother. In the other two babies, the relative affected was the father in one case and a maternal uncle in the second case.

5.4.2:2 TALIPES EQUINOVARUS

This malformation was found to be familial in two babies. In one case a maternal cousin had the same malformation and in the second baby a paternal uncle was affected.

5.4.2:3 POLYDACTYLY

One baby with polydactyly of the hands and feet had a maternal cousin with a similar malformation.

5.4.2:4 ACHONDROPLASIA

This malformation is caused by a dominant gene. One baby was born with this malformation and according to the mother the paternal grandmother of this baby had similar features. There was no other relative born to this grandmother with the same abnormality.

5.4.3: ENVIROMENTAL FACTORS

5.4.3.1: CONGENITAL RUBELLA SYNDROME

One baby was born with clinical features of congenital rubella. There was no histroy of rubella infection during the first trimester of pregnancy. The baby had bilateral congenital cataracts, microcephally, a congenital heart disease and was small for dates.

5.4.3.2 CONGENITAL POSTURAL DEFORMITIES

3 babies had congenital postural deformities. 1 baby with bilateral talipes equinovarus was a breach delivery. 2 babies with genu recarvatum, bilateral talipes equinovarus and contractures at the elbow and wrist joints were cases of extra-uterine pregnancies.

5.4.3.3 EFFECTS OF DRUGS, X-RAYS, SMOKING, ALCOHOLISM AND CONTRACEPTIVE USE

It was found that all mothers in the study and control groups had received chloroquine, folic acid and ferrous sulphate during pregnancy. Only one mother who had given birth to a child with Prune Belly Syndrome (Plate 16) had taken Stemetil and Vitamin B Complex during the first trimester of pregnancy to stop vomiting. The rest had not taken any other drugs, even local herbs.
None of the mothers in the study and control groups had been exposed to x-rays during pregnancy and none had been on the contraceptive pill before or after conception. History of smoking and alcoholism was not given by any of the mothers.

5.5 SOCIAL ASPECTS OF CONGENITAL MALFORMATIONS

The maternal reaction, knowledge about the cause of congenital malformations and social attitudes were also studied.

5.5.1 MATERNAL REACTIONS

All mothers of malformed babies showed good co-operation during the interview and expressed freely their feelings and reactions towards the malformed baby. All of the mothers of babies with obvious malformations had been told and shown the malformation immediately after birth.

Table XI shows the type of reaction in mothers of babies with major congenital malformations.

TABLE XI

TYPES OF REACTIONS IN MOTHERS OF MALFORMED BABIES

REACTION	NO.	%
Shock	74	74.5
Sadness	52	53.1
No reaction	24	24.5
Denial	7	7.1
Anxiety	7	7.1
Shame	3	3.1
Fear	2	2.0
Rejection	1	1.0
Laughter	1	1.0

5.5.2. KNOWLEDGE ABOUT THE CAUSES OF THE MALFORMATION

40.8 percent of the mothers had no explanation for the cause of the malformation. 25.5 percent did not know the cause but believed that it was God's wish that the baby was born abnormal. 33.6 percent had explanations for the cause of the malformation and these are shown in Table XII.

TABLE XII

EXPLANATIONS GIVEN BY MOTHERS ABOUT THE CAUSE OF THE MALFORMATION

CASE NO.	MALFORMATION	EXPLANATION
16	Spinabifida cystica	Eaten hot food must have burnt baby's back.
19	Spinabifida cystica	Hurt on the back during the last month of pregnancy.
53	Spinabifida cystica	"Bad eye" from sister in law because not in good terms.
12	Talipes	Abnormal position in the womb twisted the foot and hence presented out first.
51	Talipes	Foot came out first.
48	Strawberry nevus on face	Wiped the face during eclipse of the moon.
46	Collodion Skin and Microcephally	Excessive abdominal heat felt during pregnancy burnt the baby.
31	Congenital contracture of limbs	Baby outside womb, not enough space.
32	Congenital Contractures.	Lacke of space outside womb.

TABLE XII (Contd)

CASE NO.	MALFORMATION	EXPLANATION
34	Contracture of Limbs, Talipes	Baby presented with buttocks.
83	Spinabifida	Evil spirits.
58	Prune Belly	The strong drug given to stop vomiting and illnesses during early pregnancy.
36	Multiple Malformations	Illness during pregnancy.
15	Microcephally	Big ears are like his father's.
26	Amputated fingers, genu recarvatum	Mother changed positions several times while asleep.
90	Absent radius	Bad relation with husband, unwanted pregnancy.
30	Spinabifida	Trauma in the womb.

8 mothers said the malformation was hereditary. These were 2 mothers of babies with bilateral talipes equinovarus, 3 mothers of babies with harelip and cleft palate, polydactyly 2 mothers and achrondroplasia one mother.
4 mothers of babies with different types of malformations said the malformation probably came from the father's family and 3 said it was because of a bad omen.

5.5.3 SOCIAL ATTITUDES

All mothers of babies with obvious malformations except one who had been born in hospital said they would have brought the child to hospital if delivery had taken place at home. The mother of the baby with Cornelia de Lange Syndrome (Plate 19) said she would not have brought the baby to hospital since nothing could be done to correct the malformation and the baby was however going to die.

Most of the mothers of babies with correctable malformations were anxious to have their babies treated early and did not neglect the babies.

Mothers of babies with harelip and cleft palate described their babies as difficult to bring up because of the feeding problems. All the eleven mothers were not producing enough breastmilk and therefore had to supplement the breastmilk with powdered milk which was expensive and difficult to obtain. This caused financial burden to the family.

All babies with spinabifida cystica in the study had not been repaired after birth so the mothers complained of difficulty in handling the baby because of fear of inflicting pain on the defect. Because of the paralysis of the legs and

the incontinence of feaces, the mothers said they were sympathetic because the babies were going to lead miserable lives. One mother complained that she could not go to to her farm because of fear of leaving her abnormal child with other children.

Since the birth of an abnormal child may be associated with traditional beliefs and superstitions, an attempt was made to find out how many mothers had seekded opinion or treatment from traditional healers after discharge from hospital. Only one mother had visited a traditional healer. This particular mother of a baby with harelip and cleft palate wanted to protect her baby from "bad eyes" since all her other children had died during early childhood from unknown diseases.

Two mothers complained of unwanted reactions from their husbands. These mothers were blamed by their husbands for the cause of malformation.

None of the mothers complained of unwanted reactions from other family members. They were said to be sympathetic and encouraged the mother to bring the child to hospital for treatment.

Only one mother was discouraged by her relatives that the cleft palate could not be repaired. However, she did not loose hope and was anxious to have her baby repaired.

An attempt was also made to see how many mothers feared giving birth to another child after birth of a malformed baby. Three mothers said they were not planning to have another child and the reasons are shown in Table XIII. It shows that only 2 mothers feared giving birth to another child.

TABLE XIII

REASONS GIVEN BY 3 MOTHERS WHO DID NOT WANT MORE CHILDREN

MALFORMATION IN BABY	REASON	NO.
Talipes equinovarus	Completed family	1
Bilateral harelip and cleft palate	Fear of recurrence of same defect. Elder brother similarly affected.	1
Prune Belly Syndrome	Fear of recurrence of same abnormality.	1
T O T A L		3

6. DISCUSSION

In this study ninety eight babies with major congenital malformations who had been admitted in the Muhimbili neonatal ward were studied. The male to female ratio was 1:1. Fifty of those babies were born at Muhimbili Medical Centre and thirty one were referred from the other three maternity hospitals in the city. Seventeen had been born at home or on the way to hospital.

The birth rate in the three government maternity hospitals in the city is equally high but 50 percent of the babies with malformations were admitted from the Muhimbili labour ward. This may be a reflexion of the highly qualified staff at the consultant hospital since many babies with malformations are likely to be identified after delivery. Another factor is that Muhimbili caters for mothers with high risk or abnormal pregnancies. Because of the pressure of beds, mothers and their babies are usually discharged within 24-48 hours of delivery and therefore some cases may have been missed. This might also have accounted for the fewer cases from the two government maternity hospitals.

During the period of study, 27,202 babies were born alive and 98 had major malformations. Assuming that all babies with major malformations were admitted, it appears that Dar es Salaam

has a low incidence of major congenital malformations. Sukhani and other (1977) examined 17,470 babies including stillborns and found that 114 had major malformations, 91 of whom were live born infants.

6.1 PATTERN OF MAJOR CONGENITAL MALFORMATIONS

In this study, malformations of the musculo-skeletal system were the commonest and accounted for 31.6 percent of all the malformations.
Similar observations were reported by Sharma and Co-workers (1972) and by Khan (1965).

Talipes equinovarus is one of the most frequent malformation noticeable at birth. McIntosh and collegues (1954) in New York found an incidence of 5 per thousand births. This figure included live and stillborn infants with and without associated malformations. Steward (1951) noticed great differences among the various ethnic groups of Hawaii. Clubfoot was very frequent among the natives Hawaii and much rarer among the Japanese. Talipes equinovarus was the most frequent malformation of the musculo-skeletal system in the present study. The frequency of this malformation of 0.55 per thousand is lower than that reported by Khan (1965) in Nairobi where it was found to be 1.7 per thousand births. However, Simpkiss and Lowe (1961) in

Kampala reported only one case of talipes equinovarus in their series of 112 patients with congenital malformations.

The next frequent malformation affecting the musculo-skeletal system were the transverse limb reduction malformations. These were mainly the amputations of the fingers and, or toes. In the series by Sukhani and others (1977) in Lusaka, only 2 babies out of 91 live born babies with major malformations had hypoplastic hand or foot.

Polydactyly which is any duplication of one or several digits is a rather frequent malformation. In the mildest form, the extra finger is only a cutaneous appendage. In this study, such cases were regarded as minor malformations and were therefore excluded. The prevalence and incidence figures of this malformation are not uniform because of the variation in geographic areas and races. Polydactyly occured in Boston caucasian children at a rate of 1 in 1,284 but in negro children it was 1 in 117 (Altemus et al, 1965). In Kampala, Uganda, the incidence was 1.4 per thousand live births (Simpkiss and Lowe, 1961). The three cases that were seen in this study included one baby with a bifid thumb and two who each had six and seven digits on both hands and feet.

Major central nervous system malformations which include

anencephally, spinabifida cystica, microcephally, and encephalocoele were seen in this study.

There is considerable geographical variation in the incidence of central nervous system malformations. Highest incidence figures come from Western Europe and especially Britain and the communities which have emigrated from these areas, while negro and mongolian populations seem to be less affected (Penrose, 1957, Searle, 1959 and Stevenson et al, 1966). Sukhani and co-workers (1977) found that central nervous system malformations were the most frequent among the major malformations. Similar findings were reported by Leck and others (1968) and by Mathur and collegues (1975). In the present study, central nervous system malformations were the second commonest.

Penrose (1957) pointed out that anencephally is relatively uncommon in people of African origin and this is evident in this study where no case of anencephally was seen in live born babies. Great variations in the incidence of anencephally have been reported from different areas. In a world-wide study, the frequency of anencephally was extremely high in hospitals of Belfast and Alexandria, Egypt and extremely low in Bogota, Colombia and Ljubljana, Yugoslavia (Stevenson and others 1966). In Belfast, an incidence of 6.7 per thousand births, a figure was reported by Penrose (1957).

Low incidence figures are reported in studies in Africa, for example Simpkiss and Lowe (1961) did not see anencephally in 2,068 African newborn infants. Khan (1965) reported an incidence of 1 per thousand births, a figure which is higher than that reported by Stevenson and others (1966), Sukhani and colleagues (1977) and Simpkiss and Lowe (1961).

Spina bifida cystica is the commonest malformation of the central nervous system and incidence figures vary in different parts of the world. In a world-wide review, the rate was found to be approximately 0.5 per thousand births (Stevenson et al, 1966). In the present study, spina bifida cystica was the commonest malformation of the central nervous system and accounted for 70.6 percent of the malformations. Okeahialam (1974) reported only 4 cases of spina bifida cystica (including occipital meningocoele) from a study of 6 months duration in Dar es Salaam. Shija (1977) found that spina bifida was one of the commonest major congenital malformation requiring surgical treatment.

The other major malformation affecting the central nervous system is hydrocephalus.

In a comparative study of hospital incidences of congenital malformations Shija (1981) reported a low frequency of hydrocephalus and spina bifida in Dar es Salaam and the Coast Region. In this study, no case of hydrocephalus was seen.

Malformations of the gastro-intestinal system were found to be as frequent as that of the central nervous system. Similar observations were reported by Sukhani and co-workers (1977) whereby by the incidence of central nervous system and gastro-intestinal malformations were 0.57 per thousand births. Cleft lip and palate accounted for 68.8 per cent of the malformations of the gastro-intestinal system. Stevenson and co-workers (1966) in a World Health Organization study noted that the incidence of cleft-palate and cleft lip was lowest in people of African origin and is highest in Asiatic people particularly in Japanese and Chinese. The estimated incidence in the current study is about 0.4 per thousand live births. It is higher than that estimated by Norman (1971) which is 0.3 per thousand births. Lesi (1968) reported an incidence of 0.4 per thousand births in Lagos, Nigeria. Though stillborns were included, his results are similar to that of the present study. Imperforate anus comprises nearly one-quarter of the malformations of the gastro-intestinal tract requiring surgical intervention in the newborn period (Lloyd, 1964). As a single defect 5 such cases were seen in this study which accounted for 31.5 percent of the gastro-intestinal malformations.

The estimated incidence in this study of 0.18 per thousand live births is similar to the over-all incidence of 0.17/1000 reported by Stevenson and co-workers (1966). These workers reported the lowest incidence of 0.09 per thousand single births from Johannesburg and the highest, o.56 per thousand births from Kualar Lumpar.

There are three autosomal trisomy syndromes which appear to be specific and well documented. These are Down's syndrome, Edward's syndrome and Patau's syndrome (Trisomy 13-15), all were seen in the present study. Down's syndrome with an incidence of about one per five thousand live births in the commonest of all chromosomal disorders (Taylor, 1974). The incidence of this syndrome in Dar es Salaam is not known but 6 cases out of 98 malformed babies were seen. When reviewing the analysis of one-hundred and eleven karyotypes in Dar es Salaam, Mgone and Lembeli (1982) found that it was the commonest chromosomal anomally. The physical features of African children with Down's syndrome in Dar es Salaam as reported by Mgone (1982) show no major differences when compared to those of other ethnic groups. However, 5 of the six Down's syndrome babies in this study had not been recognized immediately after birth. The sixth baby was just referred because she looked abnormally short. This shows the low standard of recognition of this syndrome during the newborn period in our set up.

Therefore, the total cases that were seen in the present study do not reflect the true frequency of the syndrome.

Next to Mongolism, Edward's syndrome is the most commonly encountered autosomal trisomic syndrome. It was first described by Edwards and co-workers (1960). Combining previous collected data, Taylor (1968) made a minimum estimate of the incidence at one per 6766 live births. The same number of cases of Trisomy 18 as those of Down's syndrome were seen in this study. This could be due to the fact that such babies were admitted because of the abnormalities that are obvious in this syndrome. Trisomy 13-15 is not as common as Down's syndrome or Trisomy 18 and only 3 cases were seen in the study. Patau and collegues (1961) first identified an additional chromosome in Group D (13-15) in child with multiple congenital anomalies. These anomalies consisted of cleft lip and palate, polydactyly, microphthalmia, simian palmer creases, retroflexiable thumbs and a ventricular septal defect. Further more cases where then published in literature (Thermal et al 1961, Ellis and Marwood 1961 and Alkins and Rosenthal, 1961). Thus this established it as a specific recognizable clinical syndrome.

No chromosomal studies were done in this study but the three cases that were seen had the clinical features that have been reported by Taylor (1968).

Beckwith-Wieldman syndrome, a syndrome characterized by macroglossia, omphalocoele, visceromegally and sometime neonatal hypoglycaemia was seen in three babies. Fillipi and McKusik (1970) reported on 2 cases and reviewed the literature on this syndrome. Until 1970, together with their 2 cases, 49 had been reported and all were caucasians with the exception of one negro. Hamel (1981) reported 3 babies with this syndrome in Muhimbili, Dar es Salaam. From the number of cases reported in literature, it appears that the syndrome is rare and occurs sporadically.

6.2. RARE MALFORMATIONS

While most of the malformations that were seen in the present study are not uncommon, three rare malformations were identified.

6.2.1 MULTIPLE ANOMALY SYNDROME

This unknown type of malformation consisting of an imperforate anus and abnormalities of the musculo- skeletal system is worth reporting. This was a male baby who was born at the gestational age of 34 weeks. He was the 6th born and there was no history of drug intake, x-ray exposure, rubella infection, smoking and alcohol intake during pregnancy. Labour was uneventful and the newborn weighed 1.530 kilograms with a length of 40 centimeters and an occipital frontal circumference of 29 centimeters.

The baby died after 36 hours and possibly had other lethal internal malformations which could have been identified if autopsy had been done.

6.2.2 COLLODION BABY AND CHROMOSOMAL DISORDER

The word collodion is a descriptive term of a skin which appears varnished. Much confusion has been caused by attempting to limit the diagnosis of collodion baby to a particular entity (Well, 1966). A collodion-like covering of the skin is a rare condition. Fisher and McKee (1954) reported two cases of collodion skin of the newborn each of which was of different aetiology. One of the babies had lamellar desquamation of the

of the newborn and the second was a case of ichthyosis. The term collodion baby may be used to describe the appearance of certain babies in the neonatal period. Well (1966) cautioned that it may also be seen in cases of sex-linked ichthyosis, bullous and non-buluous ichthyosiform erythrodermia, and sjogren-Larsson syndrome.

The baby with a collodion skin that was seen in the present study is particularly interesting. Apart from the collodion skin, she also had other multiple abnormalities suggestive of a chromosomal disorder. This was a female child born at preterm and the delivery was normal. The baby weighed 1.530 kilograms, length was 31 cm and the occipital frontal circumference of 25 centimeters. He was the 4th born and the mother was 27 years. The parents were distant cousins and there was no family history of an abnormal skin condition. At birth the baby cried immediately but was noticed to be tightly encased in a collodion skin. The other abnormalities were as shown in Table VIII. The baby died after 2 days.

In order to categorize correctly a case of collodion baby, observation for at least the first year of life is required to exclude the ichthyotic conditions (Fisher and McKee, 1954). The baby in this study died after 2 days and therefore a question arises whether this was a case of lamellar desquamation of the

newborn or ichthyosis congenita.

Since this was a female child with a low birth weight and the parents were related, a diagnosis of lamellar ichthyosis is more likely. This condition was formerly called non-bulous congenital ichthyosiform erythrodermia. It is usually much more severe and delibilating than the vulgaries and x-linked types (Watson, 1978). It is present at birth. The new born had generalized erythema and is enveloped in a casing of thick stratum corneum which resembles collodion. Associated congenital anomalies have been described in literature. These are macrostomia, athyroidea, ankylosis of fingers and joints, ectropion of eyelids and lips, and rudimentary ear lobules (Shar et al, 1968, Craig et al 1970, Sehgal, 1967). Such anomalies suggestive of a chromosomal disorder possibly a trisomy syndrome.

In the great majority of cases ichthyosis is inherited and in Lamellar ichthyosis it is inherited as an autosomal recessive condition (Watson, 1978). The fact that the parents were distant cousins further supports the diagnosis of lamellar ichthyosis. To distinguish true ichthyosis from lamellar desquamation of the newborn history of the skin is essential. According to Fisher and McKee (1954) the skin is normal in lamellar desquamation while it is abnormal in ichthyosis (Frost and Weinstein, 1966).

6.2.3. de LANGE SYNDROME

de Lange syndrome was first described by Cornelia de Lange in 1933. Since her original description of three cases, many similar cases have been reported from different races. Among the reports are those of Jervis and Stimson (1963), Schlesinger (1963), Ptacek (1963) and Huang and collegues (1967). The syndrome is characterized by mental retardation, a distinctive face, characteristic hands and feet, defective growth and other multiple and minor malformations. There are essential and accidental features of the syndrome. McArthur and Edwards (1967) reported the essential features to be mental debility, low birth weight, brachycephally, hypertrophy of eyebrows and lashes, small size of hands and feet, close positioning of thumb and thenar eminence, low set ears and syndactylism of toes. The hirsute forehead, ogival palatal arch, hooked little finger and also humeral micromelia are possibly accidental symptoms.

The patient who has been described in this study was a male child who was born at full term after an uneventful pregnancy.
He was the first born to his mother who had been investigated for primary infertility. Birth was normal but was reported to

have hadapnoenic attacks and cyanosis immediately after birth. The birth weight was 2 kilograms, length 44 centimeters and occipital frontal circumference 30.5 centimeters, all below the 3rd centile for his age. The mother was 22 years and the father 27 years and were not related. The physical features are as shown in Table VIII and are similar to those reported by de Lange (1933), Jervis and Stimson (1963), McArthur and Edwards (1967), Pashayan and others (1969). These features are also similar to those described in the two negro children by Familant (1968) and by Ptacek and co-workers (1963). The case reported in this study also had monodactyly. It has been found that abnormalities of the arm can vary from phocomelia to a low placed thumb in an otherwise normal individual. A case with phocomelia was also seen by McArthur and Edwards (1967) in their series of 20 cases and by Ptacek and co-workers (1963) in the negro child.

Although the aetiology of de Lange Syndrome is not known, several hereditary factors have been proposed. Opitz and collegues (1964) described a series of families with multiple cases in siblings and suggested that the condition showed autosomal recessive inheritance. In 1967 McArthur and Edwards rejected the hypothesis of simple autosomal recessive inheritance

because of the rarity of familial cases and by the fact that the majority of reported cases are sporadic. A chromosomal abnormality has been suspected because of the multiple anomalies associated with this syndrome. Jervis and others (1963) described chromosomal abnormalities in 4 out of the 5 cases they studies. A child with a translocation of a chromosome was reported by Craig and Luzzati (1965). And also, one series of familial cases has been recorded by Falek and collegues (1966) in association with a translocation between chromosomes 3 and 21 or 22.

6.3. MATERNAL AGE AND PARITY

In this study, the mean maternal age of malformed babies was 23.9 years and that of the control 26.1 years. This difference however was not statistically significant. When the effect of maternal age and malformations of different systems was analysed, a highly significant relation was found in the trisomic syndromes and malformations of the musculo-skeletal system, p 0.01. There was no significant relation between parity and congenital malformations. Similar results were reported by Halevi (1967).

The effect of advanced maternal age and autosomal trisomic syndromes has been well documented. The most striking maternal

age effect is shown by Down's Syndrome, Edward's Syndrome (Trisomy 18) and Patau's Syndrome (Trisomy 13-15). In these three syndromes, the mean maternal age is raised with similar mean values (Taylor, 1968). In this study there were 30 mothers who were 30 years or more. 46 percent of these mothers gave birth to babies with trisomy syndromes. In 50 percent of these babies, the mothers were more than 35 years old.

It is known that the likelihood of chromosomal non-dysjuction increases rapidly after a maternal age of 30 years (Norman, 1971). Ageing of cells in the ovary, with consequent reduced meiotic efficiency, is a possible reason for the increased risk of chromosomally abnormal offspring with advancing age. Chromosome studies on populations suggest that even mitotic efficiency is reduced with age (Jacobs et al, 1963). Penrose (1933) showed that there was indeed a relationship of maternal age and mongolism irrespective of the number of children born. The mean maternal age in Down's syndrome has been found to be more than 30 years. Penrose (1961) reported a mean maternal age of 36.6 years. Sigler and co-workers (1965) reported that the mean maternal age of mothers of Down's syndrome in Baltimore, U.S.A. was 32.5 years. In a study that was done

in Dar es Salaa, Tanzania Mgone (1979) found that the mean maternal age in Down's syndrome births was 32.9 years. The mean maternal age in the 6 Down's syndrome babies in the present study was also found to be high, 35 years.

A maternal age effect and Edward's syndrome has also been reported. Taylor (1968) gives a mean maternal age of 31.8 years. In the series by Butler and others (1965) of 13 cases of Trisomy 18, the mean maternal age was 32.2 years. It was less than that in the review of 37 cases by Hecht and collegues (1963) in which it was reported to be 34.4 years. In this study, the effect of advanced maternal age and the birth of babies with Edward's syndrome is also seen. The mean maternal age in the 5 cases was found to be 31.5 years.

Snodgrass and co-workers (1966) analysed 7 cases of Patau's syndrome and found that the mean maternal age was 34.4 years. In a detailed study of 27 cases of Patau's syndrome and 27 cases of Edward's syndrome (Taylor, 1968) found a mean maternal age of 32.7 $\neq$ 1.8 years in the winter conceived group of Patan syndrome. Though the mean maternal age in the present study was 30 years, two of the mothers were less than thirty years old.

In this study it was found that trisomy syndromes occur more frequently in mothers of high parity but the results were not statistically significant. This is perhaps due to the fact that parity is related to maternal age.
A statistical significance was found between maternal age and the malformations of the musculo-skeletal system, $p < 0.01$. In the study it was found that these malformations were more frequent in young mothers. A significant relation with maternal age and malformations of the bones and joints was also found by Halevi (1967) at a 5 percent level. Similarly Erhardt and Frieda (1964) found a high rate of club foot among the young mothers. These workers also found that the rate of club foot was significantly higher among the first births.

Dunn (1976) reviewed the work and the evidence in support of the mechanical theory in causation of congenital postural deformities. In the Birmingham Maternity Hospital study of 1960-1961 as reviewed by this author, it was found that the first born infants were more often deformed at birth. The rate in the first born was 54 percent as against 35 percent in the multiparae, with a statistical significance p 0.001. Wynne-Davies (1964) also note the increased

tendency of the first born to exhibit various deformities of the feet. Though in the present study no statistical significance was shown between parity and congenital malformations, malformations of the musculo-skeletal system were commoner in mothers of low parity.

6.4 AETIOLOGICAL FACTORS

It is know that in majority of cases the aetiology of a congenital malformation is unknown. In the present study the possible aetiological factors could be identified in 25.5 percent of the cases. This figure is higher than that of 20 percent estimated by Dudgeon (1968).

In general three types of causes can operate, singly or in combination to produce congenital malformations: altered genes, chromosomal aberrations and environmental factors. Of the 98 cases that were seen in this study, 14.3 percent had clinically recognizable chromosomal disorders, 7 percent had familial conditions and 4 percent had conditions attributable to environmental causes. In the great majority of cases the doctor cannot hope to obtain clear proof of the participation of any aetiological factor in the individual case. Neel (1961) has estimated that genetic factors are

entirely or largely responsible for not more than 20 percent of all malformations, chromosomal aberrations less than 10 percent, leaving more than 60 percent presumed to be attributable to some other causes. In the present study it is seen that chromosomal disorders were recognized in 14.3 percent of cases a higher rate than that of 10 percent estimated by Neel and by Dudgeon (1968).

Several studies have been attempted to estimate the incidence of infants born with recognizable chromosomal abnormalities. Despite the great efforts involved in such investigations, the numbers of abnormalities discovered are still small. It seems that approximately 0.5-1 percent of infants are born with recognizable chromosomal aberrations (Sergovich et al, 1969, Walzer et al, 1969). Steward and associated (1969) have found that 4 percent of liveborn babies with recognized malformations have a chromosomal abnormality detectable in mitotic cells. The incidence of chromosomal disorders in Dar es Salaam is not known and therefore its magnitude cannot be compared to that of other places.

In the majority of cases that were seen in this study, 75 percent, the aetiological factor could not be identified. Among these were common malformations like spina bifida, cleft lip and

palate and talipes equinovarus. It is becoming increasingly evident that many malformations are caused by polygenic or multifactorial genetic predisposition which interacts with additional and unknown intra-uterine environmental factors. The interplay of both the multiple genes of the genetic inheritance and environmental factors are necessary for the occurance of the abnormality (Carter, 1968). Carter (1968, 1969) has reviewed the literature on some important malformations including spina bifida, cleft lip and palate and talipes equinovarus and has provided well supported data consistent with polygenic inheritance.

In the present study as an example of polygenic inheritance, 11 cases of cleft lip and palate were seen 3 of which had a positive family history. 7 of them were boys and 4 were girls. Such male excess has also been reported by Rank and Thomson (1960). The male excess, increased risk in relatives and the higher risk of recurrence in severe type of malformation are some of the distinguishing characteristics of polygenic inheritance (Carter, 1969). For example the risk to relatives increases with severity of the malformation from about 2.5 percent where the index patient has a unilateral cleft lip to about 6.1 percent where he or she was bilateral cleft lip and palate (Fraser, 1970). In polygenic

inheritance the risk of recurrence after two affected children is also higher in subsequent siblings of unaffected parents. This increased recurrence risk was seen in a baby boy with bilateral cleft lip and palate whose brother had the same malformation. In the case of cleft lip and palate, the risk for siblings born of unaffected parents has been shown to increase from about 4 percent after one affected child to 9 percent after two affected children (Curtis et al, 1961). Therefore, the mother of these two boys has an increased risk of giving birth to a child with a similar malformation in the next pregnancy.

Among the conditions due to a single gene inheritance that was seen in the present study is achondroplasia. In this baby, the grandmother was said to be very short but there was no other similarly affected relative and was therefore a sporadic case.

Achondroplasia is an autosomal dominant condition with fresh mutation rate as high as 90 percent (Nora and James, 1974). In such situations the mean paternal age is advanced (Murdock et al, 1970). Of the 117 cases that were analysed by Murdock and collegues, 79 percent were sporadic cases and 21 percent were familial, with one or both parents affected.

The case that was seen in this study is also sporadic since there was no other similarly affected relative.

As far as environmental factors are concerned, only 4 cases were identified one of them being rubella. Rubella as a cause of congenital malformations was first established by Gregg (1941). Since then many reports came up from different parts of the world (Dudgeon, 1967). Apart from this no other know environmental tertogen like drugs was indentified.

In conclusion, it is clear from this study that in most cases of congenital malformations the cause remains a mystery despite all the possible analysis.

6.5 SOCIAL ASPECTS OF CONGENITAL MALFORMATIONS

Most of the mothers of babies with apparent malformations had been told and shown the malformation immediately after birth. After such news nobody had discussed the problem with the mother about its nature, cause and the outcome. Therefore, most of these mothers were happy that someone was ready to discuss and answer their questions. A mother who had given birth to an abnormal child should be helped to overcome her anxiety. D'arcy (1968) interviewed 694 mothers who had given birth to babies with congenital defects. She found that many

mothers would have appreciated more attention paid to them and given opportunity to ask questions and to have their unvoiced fears allayed. They also would have preferred to know of the abnormality at the time of birth. In this study, a mother of Edward's syndrome baby complained that she was not being told the truth about her baby's eyes despite the questions she was asking. She had seen that the baby had no eyeballs but needed someone to confirm it to her. She was happy when she learned the truth. In our set up, mothers are shown their babies immediately after birth. This is a good practice since it relieves the anxiety when the mother learns the truth early in case the baby is abnormal.

6.5.1. MATERNAL REACTIONS

Despite the great variation among the babies' malformations, majority of mothers had similar initial reactions. 74 percent of the mothers said they were shocked when they saw that the baby was abnormal. 24 percent of them showed no emotional reaction and 53.2 percent were emotionally upset and cried when asked questions during the initial interview. Other reactions that were seen were anxiety, denial, rejection, shame

and laughter. Similar reactions were reported by Johns (1971) and by Drotar and co-workers (1975). Drotar and co-workers interviewed parents of 20 children with a wide range of malformations that included mongolism, congenital heart disease, cleft lip and palate. The course of parental reactions were determined which demonstrated 5 stages of parental reactions consisting of shock, denial, sadness and anger, adaptation, and re-organization.

One mother of a baby with unilateral cleft lip and palate was so much depressed that she even regretted marrying the husband. She would not have married him had she know of the possibility of giving birth to a child with a defect like his. In two cases the depression was made worse by the father's reactions. The mothers had been blamed by their husbands for the cause of the malformation. According to the father, the baby with arthrogyroposis multiplex congenita was born so because the mother came from a family of cripples. The relative he was referring to had suffered from polio during childhood. In the second case, the husband accused the wife of having taken drugs during pregnancy. It was because of the drugs that the baby was born with amputated fingers and toes. The mother was very unhappy because of the false accusation.

The tendency of parents to blame one another can bring about marital disharmony in the family. Such blame can arise because of previous disease, accident or habit, or through some misconception of heredity (Wilson, 1963). In the two cases in this study, the blames were not justified because the conditions were not caused by explanations given by the fathers.

Denial was seen in six mothers who had given birth to children with Down's syndrome. These mothers denied that the baby was abnormal when the information was revealed to them. Two such mothers became angry and even refused to come back for follow-up. Such malformation could not have been conceived during the initial stage of shock. But even then, one mother still denied to have been told that her child was abnormal when she was admitted for a chest infection 7 months later. Such reaction could be due to the fact that in Down's syndrome the abnormality may not be apparent to the mother until later when the child shows poor development.
It has been found that many parents prefer to be told of the diagnosis of Down's syndrome very early, possibly within one week (Drillien and Wilkson, 1964, and Gayton and Walker, 1974). Though an attempt was made to reveal the diagnosis

early in these mothers, such information was not accepted. The reason could be that our un-educated mothers do not know what to anticipate in a Down's syndrome child. To them, any child without an abvious structural defect at birth is normal. Denial was not seen in mothers of babies with obvious malformations like spina bifida.

The young mother of a baby with de Lange syndrome rejected her baby. She refused to breat feed the baby and verbally said she wished the child could die. This mother had secondary school education and probably her level of education was a contributing factor to such a reaction. Being young and having conceived after investigations for primary infertility, the birth of an abnormal child was a very traumatic experience to her. She was also very much depressed and cried a lot. In the 12 cases that were studied by Johns (1971) rejection was seen in a young mother of a Down's syndrome child but later the parents adapted to the situation. The mother in the present study could have been helped to adjust to the problem but she did not come back for follow-up. So what happened to this baby is unknown.

6.5.2. KNOWLEDGE

Mother's knowledge about the causes of congenital malformations was found to be minimal. Though 33.6 percent had explanations to give, the majority of them were wrong. Some of these explanations were due to beliefs or superstitions that the mothers had. Such beliefs and attitudes are not only seen in our African mothers. John (1971) in Melbourne, Australia reported many theories of causations of congenital malformations from mothers of abnormal children some of which were just beliefs or superstitions.

In this study, it was encouraging to find that in those babies who had familial conditions, the mothers had the right explanations. In this case it was easy to explain to the mother why the baby was born with the particular malformation. However, only one mother was worried about the risk of recurrence probably because her first child also had bilateral cleft lip and palate.

6.5.3 ATTITUDES

Mothers of babies with cleft lip and palate described their children as difficult to bring up. The commonest reasons for this were feeding problem and inadequate breastmilk

production. The later reason is expected since a mother who is emotionally disturbed is likely to fail to lactate. In this situation the mother can be helped by re-assurance and support from medical staff and relatives.

These mothers were very anxious to have their babies repaired and therefore tried their best to see that the baby gained weight despite and the difficulties.

Three babies with spina bifida cystica were followed up for 3 months, the rest had died. The mothers were anxious to have their babies operated on and wondered why it was being delayed. In all the three the defect had not been repaired before discharge from hospital and therefore dressing was to be continued at the dispensary until healing had taken place. The sight of the defect was distressing to the mothers and frequent hospital visits caused financial burden because of transport.

Despite the incontinence of urine and feaces these mothers accepted the situation and did not neglect their babies.

Hare et al (1966) also found that mothers of babies with spina bifida continued to love and want their babies despite their unnatural look. However, such babies imposed some stress in the family in some cases like marital disharmony, anxiety and fear of another pregnancy.

After the birth of an abnormal child one would expect the mother to fear giving birth to another child. This does not appear to be so in our African mothers. In this study, only two mothers feared giving birth to another child because of the possibility of recurrence. The rest said they were ready to give birth to another child anytime god wished. This attitude is contrally to that reported by Hare and collegues (1966) in mothers of babies with central nervous system malformations. One third of the mothers did not want more children when interviewed 1 month later and one fifth said they would be cautious. Though these proportions were much less after 6 months. atleast fear was present. To most of our mothers its the wish of God to give you a noraml or abnormal child. And as to when or what God gives you. one must accept and so to them the question of recurrence of the abnormality does not arise.

7. CONCLUSION AND RECOMMENDATIONS

From this study it has been found that the pattern of major congenital malformations as seen in Dar es Salaam is not very different from that of other places. Possible aetiological factors could be identified in 25.5 percent of cases and a significant relation was noted between advanced maternal age and the trisomy syndromes. This shows that in most cases the cause of a congenital malformation cannot be identified which is consistent with the findings of other workers.

1. Health workers should take more interest in the identification and diagnosis of congenital malformations since these are likely to be important causes of morbidity and mortality as communicable disease decrease.

2. Facilities for chromosomal studies should be made available to help in the diagnosis of chromosomal disorders.

3. There is a need to keep a birth defect registry in every maternity hospital so that any new syndrome can be indentified and reported.

4. Mothers who give birth to abnormal babies should be given special opportunity to discuss and ask questions concerning their babies before discharge from hospital. These mothers are emotionally disturbed and therefore need help from the medical staff to overcome their initial emotional reaction.

5. Health education on the cause, treatment, prognosis and recurrence risk of the malformation should be given to both the parents in case they may blame each other for the cause of the malformation.

6. Elderly pregnant mothers should be educated of the risks of giving birth to abnormal babies. They can be advised to seek for the existing professional expertise e.g. family planning and prenatal diagnosis for possible continuation or termination of pregnancy.

8. ACKNOWLEDGEMENTS

I must extend my sincere thanks to Dr. A. Yohani for his initial encouragement and supervision and for reading the whole manuscript.

I am indepted to the staff of the neonatal ward Muhimbili Medical Centre and the doctors and nurses from the other maternity hospitals without whose co-operation the study would not have been successful. I must also thank the staff of the Faculty of Medicine Illustration Unit, Mr. A.N. Kahangi and C. Ngowi of the Health Education Unit for helping me with the illustrations.

It would also be a crime not to extend my thanks to those unfortunate babies and their mothers. Lastly but not the least I must thank Miss Mwamvita El-Maamry and my sister Mary Bakuname who typed so carefully and tirelessly the whole manuscript.