EDITORIAL

CONGENITAL MALFORMATIONS

Congenital Malformations may be defined as abnormalities of structure present at birth and attributable to faulty development. The estimation of total incidence depends upon inclusion or exclusion of malformed still births, the care with which children are examined, the period for which children are followed up and whether domiciliary births are also included. The total incidence of congenital malformations has been shown to be probably lower in Africans in comparison to other communities. However the incidence of individual malformations differ widely in different races of man.

The role of congenital malformations in the causation of diseases of children is difficult to evaluate, as some defects are easily recognized at birth while others may go unnoticed for years. The major factors which cause congenital malformations are genetic, chromosomal aberrations, adverse intrauterine environment, infectious agents, drugs and chemicals. The variation by Socio-economic class and evidence of seasonal variation of neural tube defects have been studied. It is doutful except possibly for harelip, cleft palate and polydactyly (Khan, 1966) any variation by group can be substantiated. In a W.H.O study (Stevenson, & Johnson & Stewart, 1966) over 24 centres all over the world significant associations were found between consanguinity and neural tube defects.

Congenital malformations which are predetermined at birth either by the genotype of the foetus or by ill-understood environmental influences acting in the uterus in the first 10 weeks of pregnancy continue to contribute significantly to perinatal mortality There seems to be no possibility in the forseeable future of the prevention of these defects. It is useful to record the incidence of birth defects in local communities periodically and to collect data for prospective evaluation in future.

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Major Congenital Malformations in Neonates at U.T.H. Lusaka Zambia

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SUMMARY

The incidence of major congenital malformations in U.T.H., Lusaka during 1976 is reported. The incidence of some common major malformations are compared with the other series. Central nervous system and Alimentry system malformations were most frequent. The relative low incidence of anencephaly and cleft lip and palate in African newborns is confirmed.

INTRODUCTION

The purpose of this series of congenital malformations is to show the incidence of major congenital malformations in U.T.H. Lusaka, and to compare the incidences of some of common major malformations with other series.

MATERIAL AND METHODS

The total number of deliveries at U.T.H., Lusaka from January, 1976 to December, 1976 was taken from the records of labour ward, department of Obstetrics and Gynaecology. Major malformations were recorded in infants admitted to the Neonatal Unit and those in post-natal wards who were examined routinely. As the pressure on available maternity beds is great, mothers and their babies are discharged within 24—48 hours of delivery, hence only newborns with major malformations apparent at or soon after birth have been included in this series. A major malformation is one severe enough to cause death or significant handicap (Nelson, and Forfor, 1969). Very few autopsies were performed in both still births and those who died in the Neonatal Unit, as consent was often refused. Radiological investigations were also performed whenever indicated. We have included only those heart malformations which were detected at autopsy and excluded other heart malformations and malformations like pyloric stenosis which could not be detected at birth. We have also excluded minor malformations like talepes, polydactyly, nevi, hemengiomas etc. Multiple malformations in babies have been counted more than once while taking the incidence of different systems affected. For example, tracheoesophageal fistula was counted both in the Gastrointestinal and Respiratory malformations.

RESULTS

Of the total 17470 deliveries (still births included) in year 1976, 114 infants had major malformations giving an incidence of about 6.5 per thousand births (Table 1).

Out of 114 infants, 91 were live born infants with major malformations and 23 were still born infants with major malformations (Table 1). Eighty two newborns had single malformation and 32 had multiple malformations giving an incidence of 4.5 and 1.8 per thousand respectively (Table 1). The incidence of major malformations affecting different systems are shown in Table IIa — h. Table III shows the multiple malformations in live born infants and Table IV shows the multiple malformations in still births. Table V shows the frequency of major malformations in different systems.

DISCUSSION

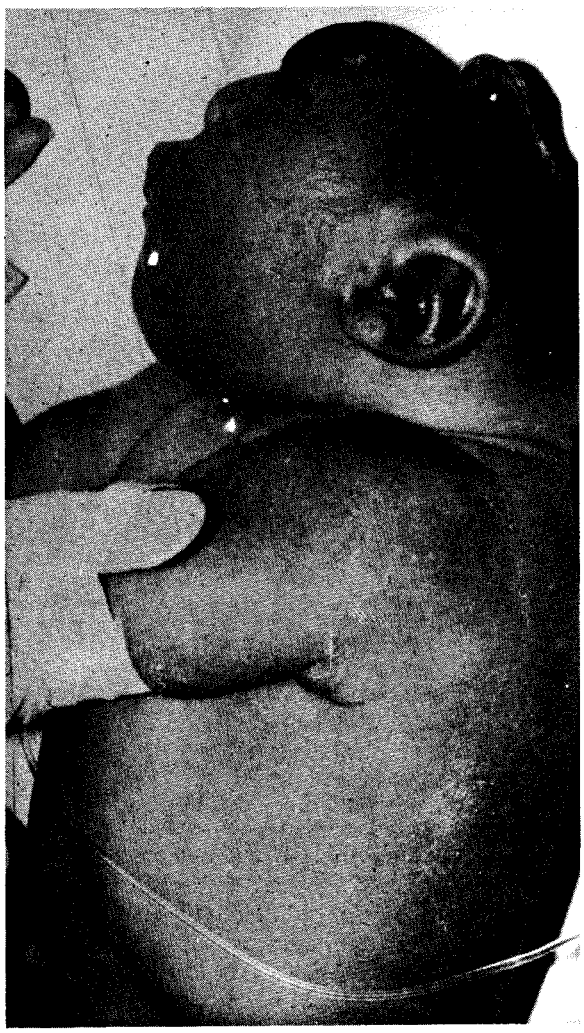
In the present series the incidence of major congenital malformations is 6.5 per 1000 births of hospital cases. This figure is low as compared to some Other reported series, for example that of Carter, 1950 (England) and Mathur et al, 1975 (India), the incidence being 14.7 per thousand births in both series. It is comparable to the incidence of 6.7 per 1000 births of hospital cases reported by Hegnauer, 1951 (Germany). However the incidence in our series is slightly higher than that of reported by Lesi, 1968 from Nigeria where the incidence was about 4.0 per 1000 births. The incidence of congenital malforma-

TABLE I

Incidence of major congenital malformations in U.T.H. Lusaka in year 1976.

	Percentage	Incidence per 1000 births
Total deliveries – 17,470	–	–
Total still births – 440	–	–
Total infants with major malformations – 114	0.65	6.5
Total live born infants with major malformations – 91	0.53	5.3
Still born infants with major congenital malformations – 23	5.22	52.2
Total infants with major single malformation – 82	0.46	4.6

FIG. I



Anencephaly

tions in different series is handicapped by the fact that no two series are made up in the same way (Neil, 1958). To avoid such handicap it is better to compare the incidence of particular malformations.

Central nervous system malformations were most frequent among major malformations (Table V). This is similar to observation of other workers (McKeown and Record 1960, Leck et al 1968 and Mathur et al 1975). The incidence of anencephaly in our series is 0.57 per 1000 births (Table IIa), which is low as compared to other reported series from Europe, U.S.A. and Asia. McKeown et al, 1960 showed incidence of 2.0 per 1000 births in Birmingham and 1.6 per 1000 births in New York. Mathur et al, 1975 showed the incidence 3.8 per thousand births in Hyderabad (India) in small series of 1016 consecutive births. Penrose, 1957 pointed out that anencephaly is relatively uncommon in peoples of African origin. Low frequencies have been reported in Pretoria where mothers were Bantu (0.5 per 1000 births) and Kampala (0.41 per 1000 births) by Stevenson et al, 1966). Slightly higher incidence of 1.0 per thousand births has been reported from Nairobi by Khan, (1965).

The incidence of spina bifida cystica (mostly meningomyelocoeles) in our series is 0.57 per 1000 births similar to that of anencephaly (Table IIa). Carter, 1967 stated that the incidence of spina bifida cystica tends to follow that of anencephaly, the same is true in our series. However Lesi, 1968 showed low incidence of spina bifida cystica (0.2 per thousand births) in relation to that of anencephaly (0.8 per thousand births) in Lagos. Okeahialam, (1974) reported only 4 cases of spina bifida cystica (including occipital meningomyelocoele) from a study of 6 months in Dar es Salaam, and one case of anencephaly. Higher incidences of spina bifida cystica have been reported in Birmingham (2.5 per 1000 births) and Sweden (1.1

TABLE IIa

Frequency of Major Malformations of different systems.

System affected	Type of malformation	Total No.	Live born	Still born	Incidence per 1000 births.
Central Nervous System	Anencephaly	10	3	7	0.57
	Hydrocephaly	10	5	5	0.57
	Spina Bifida cystica	10	10	—	0.57
	Encephalocoele	2	1	1	0.11
	Multiple brain cysts	1	1	—	0.05
	Diastematomyelia	1	1	—	0.05

TABLE IIb

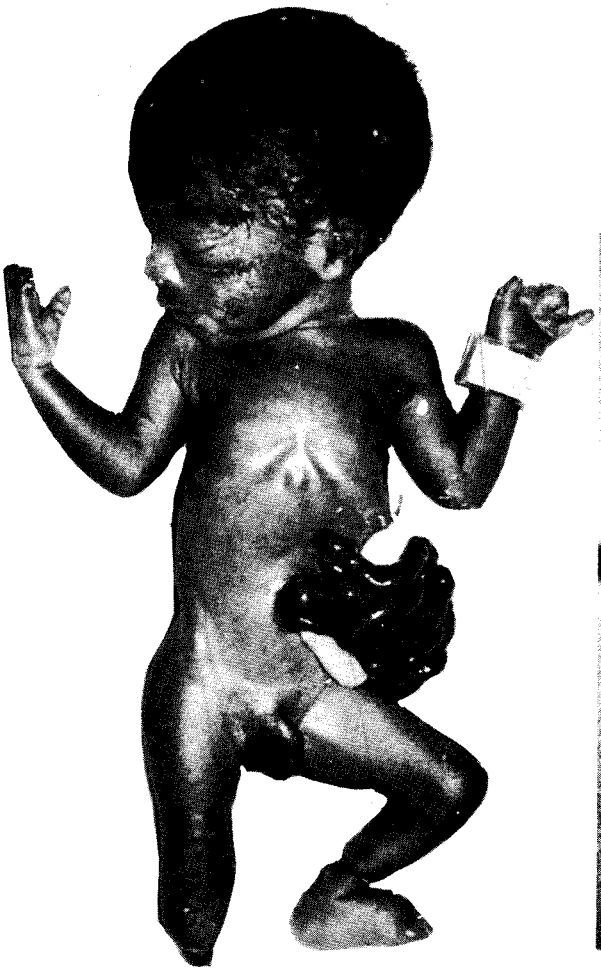
System affected	Type of malformation	Total No.	Live born	Still born	Incidence per 1000 births.
Alimentary System	Cleft lip and cleft palate	10	7	3	0.57
	Oesophageal atresia	4	4	—	0.22
	Duodenal atresia	1	1	—	0.05
	Jejunal atresia	1	1	—	0.05
	Ileal atresia	1	1	—	0.05
	Biliary duct atresia	1	1	—	0.05
	Exomphalos	8	6	2	0.45
	Imperforated anus	7	7	—	0.40

to 1.5 per 1000 births) by McKeown and Record, 1960.

The incidence of Hydrocephalus alone and with spina bifida cystica is 0.57 per 1000 births (Table IIa). It is low in comparison to 0.9 per 1000 births in Birmingham (McKeown and Record 1960), but higher than that of Lagos (Lesi, 1968), where the incidence is 0.3 per 1000 births. Stevenson et al, 1966 reported mean frequency of 0.87 per 1000 births of hydrocephaly alone and with spina bifida cystica in single births from 24 centres in WHO survey. This discrepancy may be partly due to the fact that the stay of mothers is very short in our hospital and few cases may have been missed as some of them develop hydrocephalus, sometime after birth.

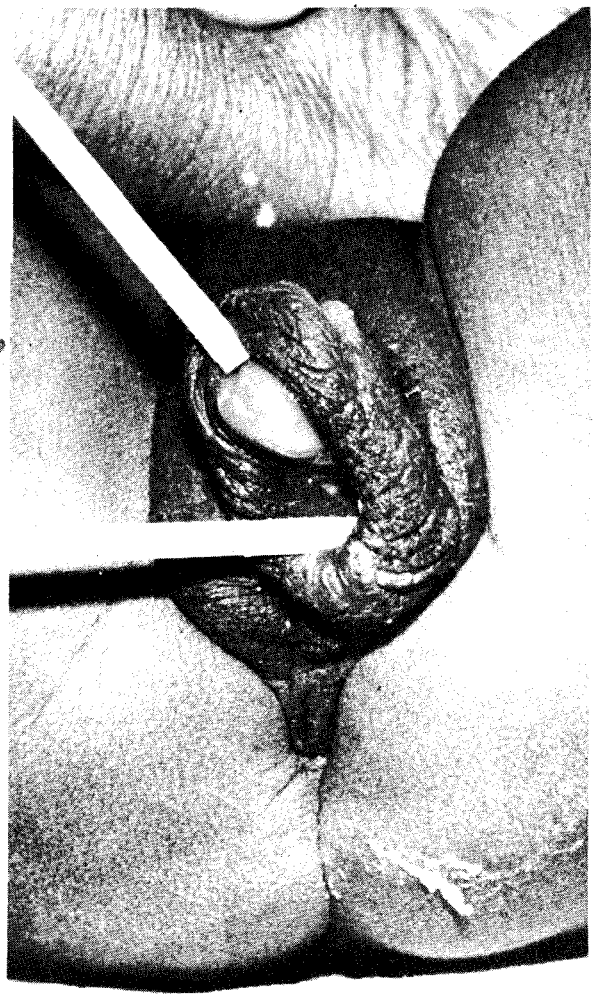
The gastro-intestinal malformations in our series were as frequent as central nervous system malformations (Table V). Mathur et al, 1975 reported incidences of 0.75 and 0.65 per 1000 births of central nervous system and gastro-intestinal system malformations respectively in their series of 1016 consecutive births from Hyderabad, India. Stevenson et al 1966 in WHO survey noted that the incidences of cleft lip and cleft palate were highest in Asiatic people particularly in

FIG. II



Exomphalos with syndactyly, constriction ring left leg and intrauterine amputation right foot.

FIG. III



Intersex

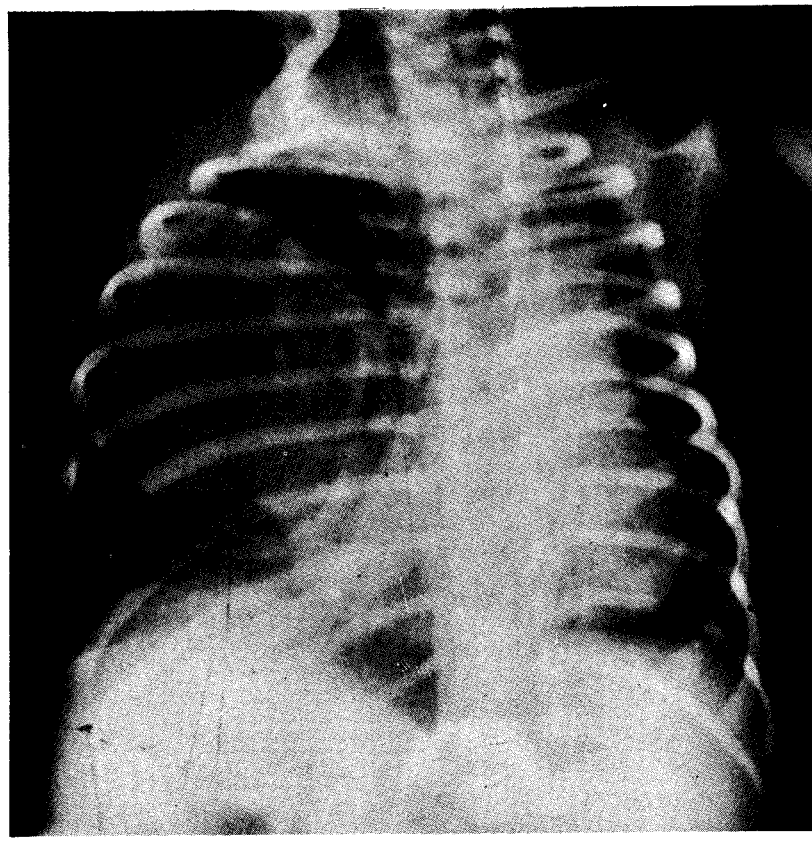
TABLE IIc

System affected	Type of malformation	Total No.	Live born	Still born	Incidence per 1000 births
Genitourinary system	Polycystic Kidneys	3	3	–	0.17
	Hydronephrosis	3	3	–	0.17
	Intersex	5	4	1	0.28
	Congenital Nephrotic syndrome	1	1	–	0.05
	Single kidney	2	1	1	0.11

TABLE IId

System affected	Type of malformation	Total No.	Live born	Still born	Incidence per 1000 births
Respiratory System	Pulmonary cysts	2	2	–	0.11
	Choanal atresia	2	2	–	0.11
	Cystic hygromaneck	1	1	–	0.05
	Tracheo-oesophageal fistula	4	4	–	0.22

FIG. IV



Pulmonary Cyst

Japanese and Chinese. The incidence of 1.4 to 1.6 per 1000 births were reported from Kuala Lumpur, Hong Kong and Singapore (Carter, 1967). The incidence is lowest in the people of African origin, about 0.3 per 1000 births according to Norman, 1971. Our incidence of 0.57 per 1000 births (Table IIb) is comparable to 0.4 per 1000 births reported in Lagos, Nigeria, (Lesi, 1968). It is higher as compared to 0.1 per 1000 births reported in Bantu from Pretoria (Carter, 1967).

The commonest congenital syndrome affecting multiple systems is Down Syndrome, the incidence of which in our series is 0.45 per 1000 births (Table IIh). It is low in comparison to 1.5 to 2.0 per 1000 births in North America, North West Europe and Australia (Norman, 1971). Stevenson et al, 1966 indicated that over most of Europe the frequency at birth appears to be between 1 and 2 per 1000 births. Few cases of Down Syndrome might have been missed as babies

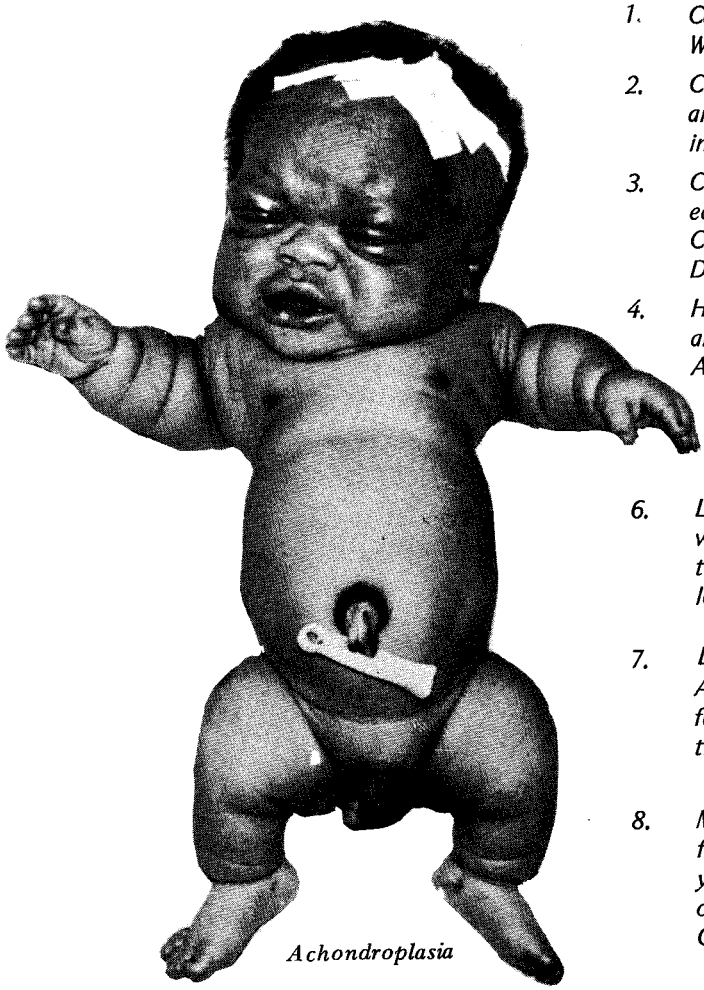
TABLE IIe

System affected	Type of Malformation	Total No.	Live born	Still born	Incidence per 1000 births
Cardio-vascular system	Ventricular septal defect	3	3	–	0.17
	Atrial septal defect	1	1	–	0.05
	Patent ductus Arteriosus	2	2	–	0.11
	Pulmonary stenosis	1	1	–	0.05
	Tricuspid atresia	1	1	–	0.05
	Hypoplastic Rt ventricle	1	1	–	0.05
	Single Ventricle	1	–	1	0.05

TABLE II f

System affected	Type of Malformation	Total No.	Live born	Still born	Incidence per 1000 births
Musculo-skeletal system	Achondroplasia	3	3	–	0.17
	Osteogenesis imperfecta	1	1	–	0.05
	Dislocation of hip	1	1	–	0.05
	Dysmelia	3	2	1	0.17
	Phocomelia	1	–	1	0.05
	Constriction rings with hypoplastic hand or foot	2	2	–	0.11
	Lobster-claw deformity	1	1	–	0.05
	Micrognathia	2	1	1	0.11

FIG. V



Achondroplasia

were discharged before being examined by us. Stevenson et al 1966 also pointed out, from their experiences over a range of hospitals, that as many as a quarter of all infants with Down Syndrome may not be recognised until after the infants have left the hospital.

The majority of cases with major malformations die during the Neonatal period or are still births. The higher incidence of major malformations among still births confirmes this (Table I).

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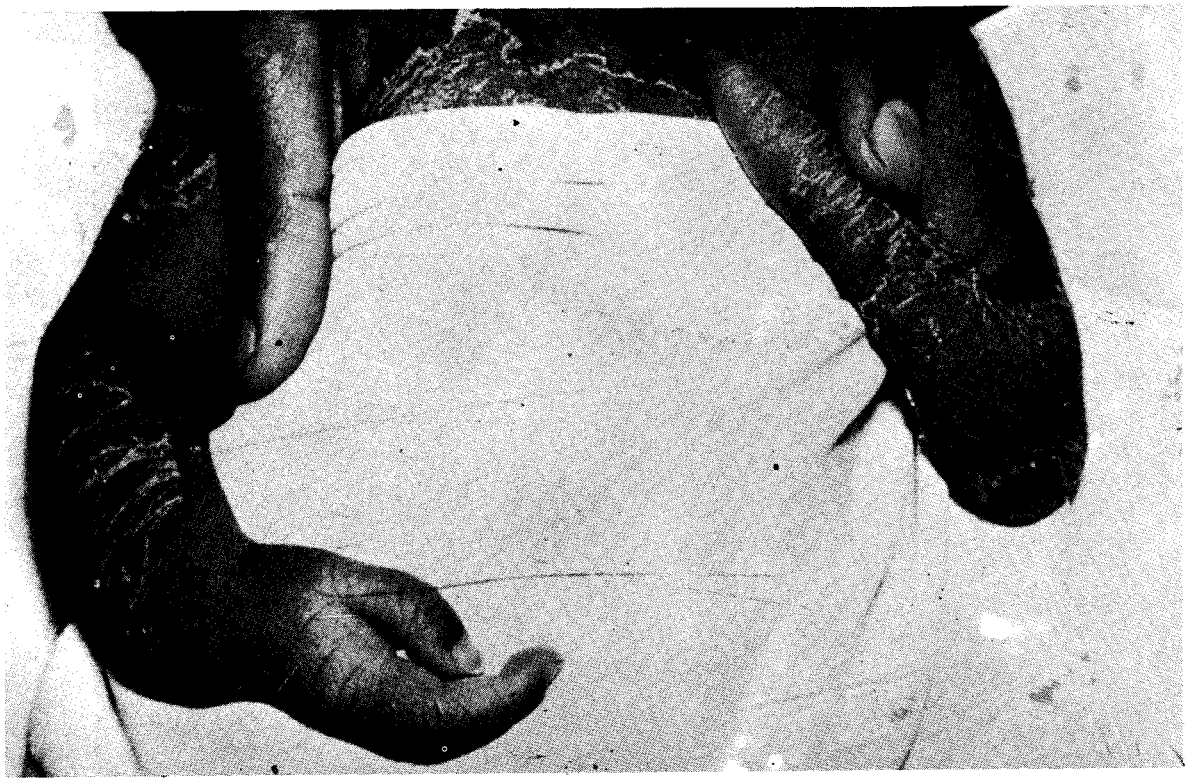
TABLE IIg

System affected	Type of anomaly	Total No.	Live born	Still born	Incidence per 1000 births
Organs of special senses	Absence of pinna	1	–	1	0.05
	Absence of one nostril	1	1	–	0.05

FIG. VI



Dysmelia

TABLE IIh

System affected	Type of anomaly	Total	Live born	Still born	Incidence of per 1000 births
Congenital Syndromes affecting multiple systems	Down Syndrome	8	7	1	0.45
	?Cri-du-chat Syndrome	1	1	–	0.05
	? Trisomy-18 Syndrome	2	2	–	0.11
	? Trisomy-13 Syndrome	2	2	–	0.11
	Sturge-Weber Syndrome	3	3	–	0.17
	Treacher-Collins Syndrome	1	1	–	0.05
	Pierre-Robin Syndrome	1	1	–	0.05
	Adrenogenital syndrome	1	1	–	0.05

FIG. VII

Down Syndrome

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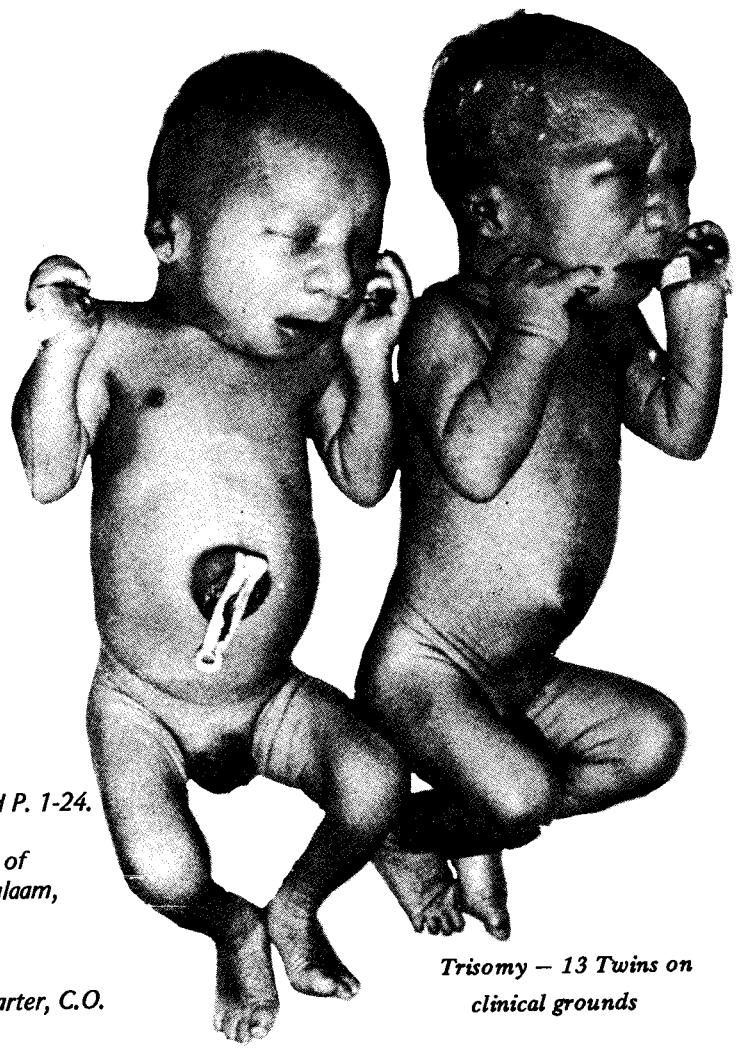
FIG. VIII



Trisomy – 13 Twins on clinical grounds

FIG. IX

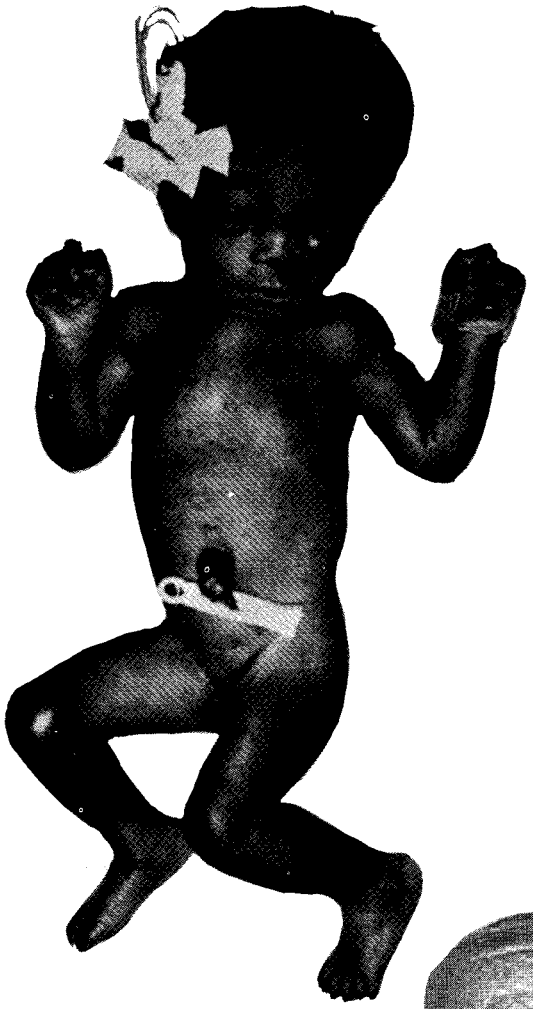


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FIG. X

Unusual position of fingers with hypoplasia of fifth fingernail in trisomy – 18 Syndrome

TABLE III
MULTIPLE MALFORMATION IN LIVE BORN
MAJOR TYPE ASSOCIATED WITH MAJOR OR-MINOR TYPE OF OTHER SYSTEMS.

Principal affected system	Type of Malformation	C.N.S.	G.I.T.	Genito-Urinary	C.V.S.	Resp.	Organs of Special senses	Musculo-Skeletal
C.N.S.	(2) Hydrocephalus	Meningo Myelocoele – myelocoele		–	–	–	–	Telepese equino-varus
	(3) Meningo-	–	–	–	–	–	–	– Do –
	myelocoeles	–	–	–	–	–	–	
	Multiple brain cysts	–	Cleft lip & cleft palate	–	–	Aplasia Rt nostril	–	– Do –
G.I.T.	Exomphalos	–	Cleft lip & Cleft palate	–	–	–	–	– Do –
	– D o –	–	Duodenal atreasia	–	–	–	–	– Do –
	– Do –	–	–	Polycystic Kidneys	–	–	–	–
	– Do –	–	–	–	–	–	–	Syndactyly with constriction ring Lt. leg and amputated Rt. foot.
	Ileal atresia	–	–	–	–	–	–	Dislocation of hip
	Imperforated anus	–	–	Rectovaginal fistula	–	–	–	
	– Do –	–	–	Hydronephrotic kidneys with perirenalcyst and intersex				
	– Do –	–	–	–	–	–	–	Talepes–equinovarus
	Bilateral cleft lip & palate	–	–	–	–	–	Bilateral absent external auditory meati	
	Cleft palate	–	–	–	–	–	–	Talepes–equinovarus
	Cleft palate	–	–	–	–	–	–	Micrognathia
	(4) Oesophageal atresia	–	–		–	Tracheoesophageal fistula	–	–
C.V.S.	Hypoplastic Rt. Ventricle							

TABLE IV
MULTIPLE MALFORMATIONS IN STILL BIRTHS – MAJOR TYPE ASSOCIATED WITH MAJOR TYPE OF OTHER SYSTEMS

Principal affected system	Type of Malformation	C.N.S.	G.I.T.	Genito urinary system	C.V.S.	Resp.	Organs of special senses	Musculoskeletal
C.N.S.	Hydrocephaly	–	–	–	–	–	–	Short neck hypoplastic extremitis
	– Do –	–	Cleft lip and cleft palate	–	–	–	–	–
	Anencephaly	–	–	–	–	–	Absent pinn (Rt)	micrognathia
C.V.S.	Single ventricle	–	–	Single kidneys	–	–	–	Bilateral Talepes equinovarus
G.I.T.	Cleft lip and cleft palate	–	–	–	–	–	–	Phocomelia

TABLE V
FREQUENCY OF MAJOR CONGENITAL MALFORMATIONS IN DIFFERENT SYSTEMS.

Systems affected	Total No.	Incidence/ 1000 births.
1. Central Nervous System	34	1.9
2. Alimentary system	33	1.8
3. Genito urinary system	14	0.8
4. Respiratory system	9	0.8
5. Cardio-vascular system	10	0.5
6. Musculo-skeletal system	16	0.9
7. Organs of special senses	2	0.1
8. Recognisable congenital syndromes affecting multiple systems	19	1.0