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Congenital malformations at birth among live-born infants in Afghanistan, a prospective study

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In a prospective study of 5,276 consecutive liveborn babies, 291 (5.5%) infants were diagnosed to have 473 congenital malformations. The incidence of major and minor malformations was 2.4 per cent and 3.1 per-cent respectively. Musculoskeletal defects accounted for 41.7 per cent of major anomalies. Among individual anomalies, congenital dislocation of hips, cleft lip ± palate, microcephaly, club feet, polydactyly, hypospadias, Down syndrome and asymmetric crying facies had a frequency greater than 1 per 1000 live-births. The incidence of neural tube defects was 3 per 1000 total births. The history of parental consanguinity was significantly higher among parents of infants with major congenital malformations as compared to unaffected control infants.

In developing countries, the developmental defects are overshadowed and often ignored due to widespread prevalence of nutritional deficiences and infectious diseases. The congenital malformations have assumed a major public health problem in the west where nutritional disorders and infectious diseases have been effectively controlled. The present study was undertaken to assess the frequency of congenital malformations at birth among live-born infants in Afghanistan. The special emphasis was placed on evaluating the correlation between history of consanguinity and incidence of congenital malformations in the offsprings.

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Materials and Methods

The consecutive live-born infants delivered at Malalaiye Maternity Hospital, Kabul during March-June 1981 were examined within 24 hours of birth for congenital malformations. A detailed physical examination was performed according to a scheme described previously[1]. The hips were examined by Ortolani's technique in all cases. The diagnosis of the malformation was based on the findings of a single examination alone because most healthy newborns were discharged from the hospital on the second day after birth. A detailed history of consanguinity among parents was obtained from all mothers. The major

congenital malformations were defined to include all those defects which caused serious structural, cosmetic and functional disability requiring surgical or medical management. The minor anomalies like nevi, skin tags, sinuses, tongue-tie, accessory nipples and Simian crease etc. were carefully looked for. Because infants with anencephaly are generally still-born, total births (live+ still births) were screened to assess the true incidence of neural tube defects.

Results

Of 5,276 live-born infants examined during the study period, 291 (5.5%) infants were identified to have a total of 473 congenital anomalies.

Major malformations

One hundred and twenty five (2.4%) infants were diagnosed to have 163 major malformations. Musculo-skeletal defects were most frequent followed by gastro-intestinal and central nervous system abnormalities (Table 1). Out of individual major anomalies, the frequency of unstable hips was found to be 6.1 per 1000 live-births. The diagnosis of subluxation of hips was based on the presence of Ortolani's click alone while infants with congenital dislocation of hips had additional clinical findings such as shortening of affected limb, abnormal perineogluteal skin crease and a dislocated femoral head without any manipulations. The other mojor anomalies with a frequency of greater than 1 per 1000 live-births, in order of their frequency, included cleft lip±palate, microcephaly, club feet, polydactyly, hypospadias, Down syndrome and asymmetric crying facies (Table 2). The diagnosis of microcephaly was based on the finding of head circumference. Unfortunately the incidence of internal systemic defects was rather low in our study because it was not possible to undertake follow up observations and examinations during the neonatal period.

Table 1 System-wise distribution of major malformations

System	No.	% of total malforma-tions	Rate per 1000 live-births
Musculo-skeletal	68	41.7	12.8
Central nervous system	24	14.7	4.5
Gastro-intestinal system*	34	20.8	6.4
Genito-urinary system	11	6.7	2.1
Cardio vascular system	1	0.6	0.2
Respiratory system	0	0	0
Chromosomal	6	3.7	1.1
Multi-system defects	30	18.4	5.7
Miscellaneous	19	11.6	3.6

*Includes infants with cleft lip ± palate and infants with congenital teeth

Out of 5,533 total births, there were 12 infants with spina bifida manifesta and five infants with anencephaly. The incidence of neural tube defects works out to be 3 per 1000 total births.

Minor malformations

Of 5,276 live births, 166 (3.1%) infants were diagnosed to have 310 minor

Table 2 Frequency of major malformations

Type of anomaly	Number	Rate per 1000 livebirths
Musculo-skeletal system		
Unstable hip	32	6.1
Congenital subluxation of hips	25	4.7
Congenital dislocation of hips	7	1.3
Club feet	10	1.9
Polydactyly	7	1.3
Syndactyly	1	0.2
Arthrogryposis	5	0.9
Limb defects	2	0.4
Aphalangia	1	0.2
Chondrodystrophy	1	0.2
Arachnodactyly	1	0.2
Chest deformity	3	0.6
Miscellaneous	5	0.9
Central nervous system		
Microcephaly	12	2.3
Spina bifida manifesta	4	0.7
Encephalocele	2	0.4
Hydrocephalus	2	0.4
Cataract	2	0.4
Anophthalmia	1	0.2
Buphthalmos	1	0.2
Gastro-intestinal system		
Cleft lip ± palate	26	4.9
Congential teeth	4	0.7
Inguinal hernia	1	0.4
Ano rectal anomaly	1	0.2
Urachus	1	0.2
Genito-urinary system		
Hypospadias	6	1.1
Ambiguous genitalia	3	0.6
Hydrocele	1	0.2
Teratoma testes	1	0.2
Cardiovascular system		
VSD	1	0.2
Chromosomal defects		
Down syndrome	6	1.1
Miscellaneous		
Facial dysmorphism*	11	2.1
Asymmetric crying	6	1.1
Pierre-Robin syndrome	1	0.2
Congenital alopecia	1	0.2

*Drysmorphism includes significant alterations in size, shape, alignment and symmetry of various components of face

anomalies (Table 3). Almost two-third of all minor anomalies were accounted for by skin nevi. The incidence of tongue-tie was around 1 per cent and its diagnosis was based on the presence of tight frenulum with a vertical groove over the tip of tongue. All the skin tags were located in front of the auricle of external ear.

Table 3 Distribution of Minor Congenital anomalies

Type of anomaly	Number	Rate per 1000 livebirths
Nevi	203	38.5
Nevus flammeus	193	
Pigmentary nevus	9	
Cavernous hemangioma	1	
Tongue-tie	50	9.5
Skin tags	17	3.2
Simian crease	15	2.8
Hooded penis	13	2.5
Accessory nipple	6	1.1
Pilonidal sinus	3	0.6
Miscellaneous	3	0.6

Malformations and parental consanguinity

Of 5,275 live-births, history of parental consanguinity was forthcoming among 1,556 (29.5%). In contrast to this, of 125 infants with major malformations, parental consanguinity was present in 48 (38.4%) cases. The difference was statistically significant ($P<0.05$).

Birth weight and gestational age

The mean birth weight of infants with major congenital malformations was 2.71 kg as compared to the overall mean birth weight of 2.91 kg among study population. Thirteen (10.4%) were low birth weight. The data suggests that there

is an excess of small-for-dates babies among malformed infants because overall incidence of low birth weight babies among the study population was 19.5%.

Discussion

The incidence of congenital malformtaions at birth among live-born babies was 5.5 per cent. It is understandable that the true incidence of anomalies would be higher because longitudinal studies of Mcintosh et al[2] revealed that merely 43 per cent of malformations were diagnosed at birth and the rest was identified subsequently during the one-year follow up. Nevertheless, the incidence of congenital malformations of 5.5 per cent is significantly higher than the figure of 3.1 per cent reported by Ghosh et al[3] in a similar study from Delhi. The incidence of major malformations of 2.4 per cent among live born infants is comparable to other studies from India.[4-8] The relatively high incidence of minor anomalies in our study is accounted for by diligent prospective search for them on a specially designed proforma.

Musculo-skeletal anomalies generally top the list of malformations in most series because they are externally visible and hence readily identified at birth. The incidence of unstable hip of 6.1 per 1000 live-births is comparable to the figure of 1-2 per cent reported by Western workers[9]. It is, however, much higher than the figure of 1.1 per 1000 live-births reported by Singh and Sharma[10] in a prospective study from North India. The diagnosis of congenital dislocation of hips can be easily missed unless all infants are examined thoroughly at birth for the stability of the hip joints.

The incidence of internal anomalies is rather low in our study due to lack of follow up. The incidence of neural tube defects of 3 per 1000 total births is lower than the reports from North India.[10-11] The incidencee of microcephaly was rather high and needs further studies to identify the etiology.

The pattern and frequency of idividual anomalies is comparable to similar studies from India. The incidence of cleft lip+palate (4.9 per 1000 live-birth) was significantly higher and there is a need to undertake correlative and analytic case studies to identify possible causal factors. The frequency of Down syndrome of 1 : 879 live-births is identical to other studies from abroad.[10-12] It is likely that infants with facial dysmorphism and multi-system defects have underlying chromosomal of multigenic defects which can be identified by karyotyping.

The religious sanction of marriage among blood-relatives is well known in Islamic countries. The history of parental consanguinity was obtained in almost one-third of the study population. The incidence of consanguinity was significantly ($p<0.05$) higher among parents of infants who had major malformations as compared to parents of those who were normal at birth. However, adverse impact of consanguinity is likely to be greater in relation to genetic defects or inborn errors of metabolism rather than visible structural defects. There is a need to undertake community surveys to identify the incidence of genetic disorders and their correlation to parental consanguinity in Afghanistan.

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References

1. Singh M : Early diagnosis of congenital malformations. In Care of the Newborn. Sagar publications, New Delhi. Second Edition 1979, p 59
2. McIntosh R, Merritt KK, Richards MR, Samules MH, Bellows MT : The incidence of malformations. A study of 5,964 pregnancies. Pediatrics 14 : 505, 1954
3. Ghosh S, Bali L : Congenital malformation in the newborn. Indian J Child Health. 12 : 448, 1863
4. Saifullah S, Chandra RK, Pathak IC, Dhall GI : Congenital malformations in the newborn period. A prospective longitudinal study. Indian Pediatr 4 : 251, 1967
5. Tibrewala NS, Pai PM : Congenital malformations in the newborn period. Indian Pediatr 11 : 403, 1974
6. Khanna KK, Prasad LSN : Congenital malformations in the newborn. Indian J Pediatr 34 : 63, 1967
7. Master-Notani PN, Kolah PG, Sanghvi LD : Congenital malformations in the newborn in Bombay—Part I and II. Acta Genet 18 : 97 1968
8. Mathur BC, Karan S, Vijaya Devi KK : Congenital malformations in the newborn. Indian Pediatr 12 : 179, 1975
9. Cyvin KB : Congenital dislocation of the hip joint. Acta Pediatr Scand. Supplement 263, 1977
10. Singh M and Sharma NK : Spectrum of congenital malformations in the newborn Indian J. Pediatr 47 : 239, 1980
11. Verma IC : Neural tube defects in India. In Medical Genetics in India. Vol. I. Ed Verma IC. Auroma Enterprises, Pondicherry, 1978, p 33
12. Aiyar RR, Agarwal JR : Observations on the newborn—A study of 10,000 consecutive live-births. Indian Pediatr 6 : 729, 1969