Brief Reports

Congenital Ocular Malformations at Birth

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Congenital ocular malformations are the result of defective development of ocular tissues during the intrauterine life, which can result from adverse genetic effects, environmental factors, teratogens or chromosomal anomalies in the developing embryo. The array of ocular findings associated with these developmental disorders, metabolic and systemic diseases and chromosomal anomalies is endless. The reported incidence of congenital ocular malformations varies from 10.5 per 1000 births(l) to 0.75 per 1000 births(2) depending on the population studied and method used. We analyzed the congenital eye malformations noted among 12,797 consecutive deliveries in a referral hospital in South India.

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Subjects and Methods

This prospective study was undertaken from September 1989 to December 1992. All the babies born during this period were examined for the presence of congenital anomalies of the eye. Relevant antenatal details including parity, gestational age, consanguinity, illness and drug ingestion during pregnancy were recorded.

Birth weight of babies, sex and condition at birth were also noted. Thorough physical examination was done to detect other associated anomalies. Babies born during the same period without eye malformation were compared with the cases. The data was analyzed using Chi-square test.

Results

There were 12,797 births during the study period, of which 12,337 were live births and 460 were still born. The number of babies with congenital malformations was 353 with an overall frequency of 2.76%. The malformations observed among live and still births were 308 (2.5%) and 45 (9.8%), respectively. A total of 21 eye malformations were found in 16 babies (1.64 per 1000 births). Of these, 13 were live births and 3 were still born. The difference in malformations among live and still births was not statistically significant. There were no eye malformations observed among 177 cases of multiple pregnancies, including 2 sets of triplets, during this period. Microphthalmos was the commonest eye malformation observed followed by absent eye lashes and anophthalmos. There were associated anomalies among 11 babies *(Table I).* Consanguinity of parents (n = 10) ($p < 0.01$) and lack of antenatal care (n = 13) ($p < 0.001$)

TABLE I–*Ocular and Associated Anomalies in Relation to Consanguinity*

Sl. No.	Consan-guinity	B/UB	Eye anomalies	Associated anomalies
1.	NC	UB	(Rt) Microphthalmos & (Lt) Anophthalmos	Bilateral absent thumb & Verticular septal defect
2.	NC	UB	Bilateral microphthalmia	Short limbs.
3.	NC	UB	(Rt) Microphthalmos aniridia and choroid coloboma (Lt) Iris & choroid coloboma	———
4.	C	UB	Bilateral microphthalmia, absent eye-lash	Collodion baby
5.	C	UB	Bilateral microphthalmia	Cleft lip & cleft palate polydactyly
6.	NC	UB	Bilateral microphthalmia, secondary glaucoma	———
7.	C	UB	(Rt) Microphthalmos	———
8.	C	UB	(Lt) Microphthalmos	———
9.	C	UB	Bilateral anophthalmia	Short lower limbs Normal Upper limbs
10.	C	UB	(Rt) Anohthalmos Absent left eyelid	Absent skull vault Hypoplastic right side of face & cleft palate
11.	C	UB	Bilateral cataract, microcornea	———
12.	NC	B	Dermoid cyst of (Rt) medial canthus	———
13.	NC	B	Buphthalmos	———
14.	C	B	Absent eye lashes	Collodion baby
15.	C	UB	Absent eye lashes	Collodion baby
16.	C	UB	Absent eye lashes	Collodion baby

C= Consanguineous; NC= Non-consanguineous; B= Booked; UB= Unbooked.

were significantly associated with eye malformations.

Discussion

The frequency of congenital ocular malformations in the present study was 1.64 per 1000 births as compared to 0.18 per 1000 births in Hyderabad(3) and 0.49 per 1000 births in Bombay(4). The higher rates in the present report may be related to the high incidence of consanguineous marriages, malnutrition and infections. Moreover, ours was a prospective study. A comparable figure of 1.5 per 1000 births was reported from Jamnagar(5). However, no specific eye malformation was listed in a retrospective study of congenital malformations among 10,000 babies from Ludhiana(6). The high figure of 10.5 per 1000 births reported from North India is probably due to the methodology, where authors included persistent hyaloid system as an anomaly, which is normally present in as much as 95% of preterm babies(7).

Microphthalmos was the commonest ocular anomaly noticed. Stolle *et al.* reported microphthalmos as the second commonest congenital ocular anomaly (2). There were 10 cases of microphthalmos and 6 anophthalmos among 354 congenital ocular anomalies seen in the Department of Ophthalmology(8). Microphthalmia and anophthalmia are due to defective or failure of development of optic primordium. Microphthalmos is commonly associated with other anomalies like coloboma or cataract. In the present study, one baby with microphthalmia had aniridia and coloboma of the choroid, and another had secondary glaucoma. Six babies with microphthalmia had associated extraocular malformation. The findings observed in these babies did not suggest any recognized syndrome(9). Warburg has reviewed the genetics of microphthalmos and identification of these defects are important in genetic counselling(l0). Animal experiments have shown that vitamin A deficiency in the developing embryo can produce anophthalmos, microphthalmos, and hair lip(ll). Reports are also available to indicate the relationship of maternal vitamin A deficiency and microphthalmos(12,13). Other causes of microphthalmia include intrauterine infections, chromosomal anomalies and teratogenic drugs. In the present study, 6 of these were born to consanguineous parents. The large number of these malformations seen in Pondicherry could be the result of consanguineous marriages, malnutrition and infectious diseases prevalent in the population(8).

Four babies who had absent eyelashes had congenital icthyosis, which was the cause of ocular findings in these babies. Cataract was the commonest congenital ocular anomaly noted in several studies(2,8). Often cataracts do not appear in the newborn period but at a later date. Since the present study was restricted to the newborn period, presence of congenital cataract in one baby does not reflect the true incidence.

Out of sixteen babies, eleven babies had associated abnormalities of one or more system. Burck *et al.* reported that out of 127 patients who had congenital malformations of the eye and orbit, only eleven had single ocular anomaly without associated malformations(14). These observations make it imperative that a child with congenital eye malformation should have a thorough systemic examination to detect anomalies in other systems.

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