Asymmetric crying facies and congenital anomalies

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Perlman, M., and Reisner, S. H. (1973). *Archives of Disease in Childhood*, 48, 627. **Asymmetric crying facies and congenital anomalies.** The frequency of hypoplasia of the depressor anguli oris muscle in a newborn population was 41 in 6360 (1 in 155). No adverse factors were noted in the obstetric background of affected infants and the pathogenesis of the lesion is not clear. The incidence of associated congenital anomalies detected in the newborn period was 2 in 41 (4.9%). The lesion was predominantly left-sided (83%) in this series, which contrasts with the side of the lesion in infants found in hospital populations (32%). Even allowing for the anticipated increase in incidence of other anomalies during follow-up, the prognosis appears to be more favourable than that indicated by studying series of infants ascertained by way of associated anomalies.

Congenital hypoplasia or agenesis of the depressor anguli oris muscle (DAOM), causing 'asymmetric crying facies', has recently stimulated interest owing to observed associations with other congenital anomalies (Caylor, 1969; Chantler and McEnery, 1971; Pape and Pickering, 1972). Evaluation of the significance of such associations has not been possible since the frequency of the phenomenon in the general population and the incidence of associated malformations in an unselected series have not been determined.

The results of a prospective survey of a newborn nursery population are reported here. The obstetric background and neonatal course of infants with the sign are compared with those of a control group in an attempt to detect pathogenetic factors. The incidence of other clinically detectable congenital malformations was determined in the newborn period.

Material and methods

All infants of birthweight 2000 g or more, born in this hospital between 1 September 1967 and 31 December 1969, were examined. Infants weighing less than 2000 g at birth were transferred immediately to a premature unit and are not included in this study. A total of 6360 infants were examined.

The facies were examined during crying at the end of the routine neonatal examination. Criteria for clinical diagnosis of hypoplasia of DAOM were similar to those in other publications (Hoefnagel and Penry, 1960; McHugh, Sowden, and Levitt, 1969; Nelson and Eng,

1972). Positive findings were confirmed by a second observer.

Gestational age, intrauterine growth, and the presence of other anomalies were evaluated by standard clinical methods (Lubchenco *et al.*, 1963; Usher, McLean, and Scott, 1966).

The control group of newborn infants was the same as that employed in a previous study (Reisner *et al.*, 1971).

Results

Weakness of the DAOM was found in 41 of 6360 infants examined, an incidence of 1 in 155. The lesion was on the left side in 34 infants and on the right side in 7. No other neurological findings were detected. During the study period no case of complete peripheral seventh nerve paresis was seen.

The study group was similar to the control group with respect to the following: maternal age, parity, incidence of complications of pregnancy, mode of delivery, and birth condition. Sex incidence was equal. Birthweight of the infants was between the 10th and 90th centile for gestational age in 40 infants; 1 infant was large for dates. No unusual racial incidence was noted. There were two instances of parental consanguinity.

Three mothers mentioned other cases of facial asymmetry in the family. In two (an elder sib in one case, a third cousin in the other) it appeared that the lesion was total unilateral facial paresis. In the third case, the mother reported that 3 daughters of her brother had asymmetric crying facies since birth, identical to those seen in her own daughter.

Associated findings are recorded in Table I.

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FIG.—Typical appearance of 'asymmetric crying facies' due to hypoplasia of DAOM (a) at rest, (b) crying. There is a hint of asymmetry of the mouth at rest.

TABLE I
Clinical characteristics of 41 infants with hypoplasia of DAOM and associated findings

Side of lesion	
Right	7
Left	34
Sex	
Male	21
Female	20
Family history of VII nerve paresis	2
of asymmetric crying facies	1
Consanguinity	2
Intrauterine growth retardation	0
Associated anomalies	
Anterior dislocation, knees bilateral	1
Coarctation of aorta	1

The incidence of major congenital malformations was 2 in 41.

Discussion

Weakness of the DAOM was first described by Hoefnagel and Penry (1960). Electrical studies were carried out by McHugh *et al.* (1969) and their findings were interpreted as consistent with agenesis of the muscle. Nelson and Eng (1972) have recently confirmed and added to these findings. Though electrical studies were not performed in the present series, the clinical findings conformed to the typical picture (Fig.) of hypoplasia of DAOM in each case.

The pathogenesis of the lesion is unknown. A family history of 'asymmetric crying facies' was obtained in one instance in the present series. No noxious influences during pregnancy were identified in this and other series (Caylor, 1969; Pape and Pickering, 1972).

Mechanical forces (such as shoulder pressure)

have been invoked as the cause of congenital facial paresis owing to an observed relation between the side of the lesion and the side of the obstetric position and direction of external restitution during delivery (Hepner, 1951). As in Hoefnagel's cases, the possible role of this factor could not be evaluated in the present series. Possible support for a mechanical aetiology comes from 4 cases in the series of Pape and Pickering (Cases 14 to 17 inclusive, left side). Variable combinations of malformed ear, torticollis, congenital dislocation of the hip, scoliosis, and unilateral mandibular hypoplasia were observed. These asymmetric signs are interrelated (Watson, 1971) and suggest that postural intrauterine factors may have been involved in the pathogenesis of the lesion of DAOM in these cases.

The frequency of hypoplasia of DAOM in the general population as well as the incidence of associated congenital anomalies in an unselected series have not been previously reported. The frequency in this population (1 in 155) is high, and it is therefore not surprising that the incidence of associated anomalies is considerably lower than that in infants ascertained in cardiac clinics (Caylor, 1969; Chantler and McEnery, 1971) and in a hospital population (Pape and Pickering, 1972) (Table II). Among associated anomalies reported, cardiac malformations have been disproportionately represented. Whether this is due to a real association ('cardiofacial syndrome'), or to biased sampling, cannot be established at present. As in the series reported here, the incidence of associated anomalies in infants referred to neurological and otolaryngological clinics was low (Hoefnagel and

TABLE II

Relations between method of ascertainment of DAOM hypoplasia, incidence of associated congenital anomalies, and side of lesion

Method of ascertainment	Reported major congenital anomalies		% Left-sided
	No.	%	
(1) Unselected*	2/41	4.9	83
(2) Neurological ENT (McHugh <i>et al.</i> , 1969; Hoefnagel and Penry, 1960)	0/8	0	100
(3) Hospital population (Pape and Pickering, 1972)	32/44	72.7	54.5
(4) Cardiac clinics (Caylor, 1969; Chantler and McEnery, 1971)	17/17	100	17.6

*Present series.

Penry, 1960; McHugh *et al.*, 1969; Nelson and Eng, 1972). Until the results of prospective studies of DAOM lesions become available, we believe that caution should be exercised in interpreting the finding as an index of the presence of other congenital anomalies. We are following this group of infants and feel that it is not yet justified to perform extensive radiological investigations such as skeletal surveys and intravenous pyelography in infants with hypoplasia of DAOM as an isolated finding.

The lesion in the present series as well as in those from neurological and otolaryngological clinics was predominantly left-sided. This contrasts with cases associated with congenital anomalies in which right-sided lesions predominate (Table III). These preliminary observations suggest that the frequency of associated congenital anomalies may be related to the side of the lesion.

TABLE III

Side of lesion of DAOM related to incidence of congenital anomalies*

Side of lesion	Total no.	Major anomalies	
		No.	%
Right	41	29	70
Left	69	22	32

*Derived from Caylor, 1969; Chantler and McEnery, 1971; Pape and Pickering, 1972; Hoefnagel and Penry, 1960; McHugh *et al.*, 1969; and present series.

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