Correspondence

does not permit full discussion of these families but
suffice it to say that the genetics of the various
craniosynostoses are unlikely to be as simple as
Dr Escobar might wish to think.

We agree that the mother of our patient shows many
of the features in adulthood of the other ACS syn-
dromes and in particular ACS type III. However, as
infants, both patients presented with clear-cut
trigonocephaly and lacked the soft tissue syndactyly
seen in Chotzen syndrome. Certainly neither patient
would have been recognised as a case of Chotzen
syndrome or any of the other ACS syndromes in the
newborn period. Therefore, had either of these infants
been seen for genetic counselling in the absence of a
positive family history, there would have been no
rational for counselling on the basis of an autosomal
dominant gene. At present there is no information as
to the frequency with which isolated craniosynostosis
represents new dominant mutations. We, therefore,
felt it important to report autosomal dominant
trigonocephaly and in particular to emphasise the
importance of minor acroskeletal anomalies in
distinguishing the familial cases. The importance of
these minor anomalies has been proven to be useful in
other families multiply affected by craniosynostosis
(Hunter and Rudd, 1977). Our patients are clearly
not typical examples of the Chotzen syndrome and
whether or not they represent variable expression of
the Chotzen syndrome gene, or a mutation, either
allelic or at a different locus cannot be determined
from the current state of knowledge. Observation of
the experiences of biochemical geneticists should lead
us to expect genetic heterogeneity in craniosynostosis,
with or without associated syndactyly or poly-
syndactyly.

Yours, etc,

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References

Cohen, M. M. (1975). An etiologic and nosologic overview of
craniosynostosis syndromes. Birth Defects: Original Article
Series, XI(2), 137-189.

Sagittal synostosis; its genetics and associated clinical
findings in 214 patients who lacked involvement of the
coronal suture(s). Teratology, 14, 185-194

Coronal synostosis; its familial characteristics and asso-
ciated clinical findings in 109 patients lacking bilateral
poly syndactyly or syndactyly. Teratology, in press.

Pfeiffer, R. A. (1964). Dominant erbliche Acrocepha losyndak-

Pfeiffer, R. A. (1969). Associated deformities of the head and
(Case No. 3).

Vogt, A. (1933). Dyskephalie (dysostosis craniofacialis,
maladie De Crouzon) und eine neurartige Kombi-
nation dieser Krankheit mit Syndaktylie der 4 Extremi-
taeten (Dyskephalodaktylie. Klinische Monatsblatter für
Augenheilkunde, 90, 441-454.

Risk of closed lesions in sibs of cases of open neural
tube defect

Sir,

Recent studies (Wynne Davies, 1975; Carter et al.,
1976) have shown that malformations resulting
from defective closure of the neural tube cover a
spectrum ranging from spina bifida occulta through
open meningomyelocele to anencephaly.

Families at risk for the recurrence of neural tube
defects may have a child with any degree of the
abnormality, or one with hydrocephalus alone
(Lorber and Ne, 1970). Alpha fetoprotein (AFP) in
amniotic fluid is recognised as a reliable index of
open neural tube defect in the fetus (Brock and
Sutcliffe, 1972), allowing selective termination of
potentially handicapped children.

Two cases of true closed lesions occurred in 12
recurrences in the first 140 pregnancies 'at risk' for
recurrence of neural tube defect in N.S.W., Australia.
The previous abnormality was spina bifida in 91 cases
and anencephalus in 49 cases.

Both cases described showed no evidence of
abnormality on antenatal testing (serial echograms,
serum, and amniotic fluid AFP at 15 to 16 weeks of
pregnancy). The previous sib had died after con-
servative management of severe open thoracolumbar
meningomyelocele in each instance.

The first case, a female, was noted at birth to have a
mobile swelling to the left of midline in the inner
quadrant of the buttock. There was no neurological
deficit. X-ray pictures of the spine were reported as
normal. Myelogram at 15 months showed a sacral
meningocele with tethered cord. Excision of the
lipoma, and freeing of the cord were performed and
the child is developing normally.

The second, a male, was noted at birth to have a
head circumference of 39·4 cm, which increased
rapidly to 44·3 cm at 3 weeks of age. Investigation by
air ventriculogram showed moderate hydrocephalus
with pronounced asymmetry, the left lateral ventricle
smaller than the right, displacing the septum pelluci-
dum across the midline. A large paraventricular cyst
stretched from vertex to occiput region on the left
side and the blockage was at aqueduct level. A ventri-
culoperitoneal shunt was inserted. The head circum-
ference is continuing to increase, and at 5 months he
is showing delay in developmental milestones.
The prognosis is poor; there is shunt dysfunction and mental retardation.

The 8.6% frequency for recurrence represents a biased parental risk situation. Five couples had 2 previous affected children, and 3 had 3 affected. In three instances one parent had spina bifida occulta as well as a previously born anencephalic child.

The proportion of 'closed' to 'open' lesions (1 to 6) seen in this series may show a similar false elevation.

In previous studies the proportion of sibs with anencephaly and spina bifida was 2.4% where the index case had congenital hydrocephalus (Lorber and, Ne, 1970), 4.2% where the index case had spinal dysraphism (Carter et al., 1976), 4.8% where the index case had multiple vertebral anomalies including spina bifida occulta, and 2.9% where the index case had multiple vertebral anomalies alone (Wynne Davies, 1975).

The risk for closed lesions must be included in counselling parents seeking antenatal testing, and the fact that, by present methods, they are not detectable during pregnancy, that the prognosis for completely normal development is guarded.

Yours, etc,
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References
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