SCAPHOCEPHALY, OXYCEPHALY AND HYPERTELORISM. With Reports of Cases.

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The congenital abnormalities of the head, which have been described under the names of scaphocephaly, oxycephaly and hypertelorism, are sufficiently uncommon to justify the publication of such cases when they occur. The nomenclature of these conditions has been in a state of some confusion, and the work of Park and Powers⁽¹⁾ and Greig⁽²⁾ ⁽³⁾ has been valuable as presenting a clear clinical and anatomical picture in each case.

Case I at 9 months.

The cases which are included in this study offer an opportunity of contrasting and comparing these deformities, and of considering whether the differences between them are not differences of degree rather than kind. Greig has pointed out that the most striking feature both of oxycephaly and hypertelorism, anatomically, is an abnormal shape of the sphenoid bone.

It is our intention to describe a case of scaphocephaly and a case of hereditary oxycephaly, and briefly to express our reasons for complete agreement with Greig on the question of the importance of the sphenoidal abnormality.

CASE 1. A. C., æt. 6. Female. Was brought to the Out-Patient Department of the East London Hospital for Children because of whooping cough and increasing shortness of breath. There was a history of a fortnight's illness, during which time the child had been getting steadily worse.

On examination, the curious shape of her head and the vacant, staring expression, combined with the extreme exophthalmos, gave her a most bizarre appearance. She was pale and obviously extremely ill. There was a broncho-pneumonia of the lower lobes of both lungs. No other abnormality was discovered, apart from webbing of the second and third toes of each foot.

The Head. The vault was high and the vertex was shaped like the keel of a ship, the forehead coming to a point resembling the bow, and the occiput being prominent posteriorly, simulating the stern. The highest point was in front of the anterior fontanelle, at the site of which there was a marked depression. The head was elongated antero-posteriorly, and narrowed laterally, as is shown in the photographs and by the following measurements :—

Nasion to external occipital protuberance, $13\frac{1}{2}$ ", the normal for that age being $12\frac{1}{4}$ ".

The parietal eminences were absent, the bi-parietal measurement being 11" against the normal 13".

The circumference was $19\frac{1}{2}$ ", quite normal.

Case I at 9 months.

The face was narrowed, measuring 5" from the root of one zygoma to that of the other, the normal being 6".

The palate was highly arched, and only one molar tooth was present on either side.

On transillumination the maxillary antra appeared to be non-existent. Exophthalmos was extreme. There was no nystagmus or strabismus, and the pupils reacted to light. Mr. Davenport reported that the optic discs were normal. Hearing, taste and smell were normal as far as could be ascertained under the circumstances; the child had a vacant expression and was very dirty in her habits, passing her dejecta under her. She appeared to live in a state of constant fear, crying out when approached, and it was decided that her mental state was due to chronic neglect and ill-treatment, rather than to actual mental deficiency.

Blood. There was a lymphocytosis, ascribable to the pertussis, nothing else abnormal.

Wassermann reaction. Not done.

Urine. Quite normal in every respect.

X-ray of Skull. Marked thinning of the bones of the vault. Digital markings seen. Flattened pituitary fossa, and marked under-development of both wings of the sphenoid. The appearances are those of deficient calcification. (Dr. B. Leggatt.)

Previous History. Full-term child—normal labour—is the youngest of four children, all healthy. The head condition is stated very definitely to have been present at birth. At the age of nine months she was taken to the Hospital for Sick Children, Great Ormond Street, because of the shape of the head. The following is the note of Dr. Donald Paterson, then Medical Registrar:—"Eyes protruding from the head like exophthalmic goitre. The back part of the head seems fairly round. The front portion has the appearance of having been pinched up in all directions—side to side—before backward. Child smiling—takes a great interest in surroundings, sits up and can stand if supported."

Family History. Mother had two miscarriages before the birth of this child.

Progress. She went rapidly downhill and died ten days after admission.

Post-mortem. (Dr. Temple Grey.) Done ten hours after death. The skull, on being opened, is seen to consist of the outer table only, and to be greatly thinned, being semi-transparent and in places actually deficient. There is complete absence of diploë. The skull is sutureless. The digital markings are well shown. The sphenoid was seen to be much smaller than normal, the superior orbital fissure being generally contracted to a gross degree. The orbital cavities were of normal shape, but were very small, being generally contracted. They contained a marked excess of fat. The frontal, maxillary and sphenoid regions contained no air-cells. The brain was normal, and shewed no evidence of atrophy. The lungs were the seat of an acute septico broncho-pneumonia. The kidneys, thymus and thyroid were normal. The webbing of the toes has already been mentioned.

CASE 2. D. F., æt. 8½ months. Female. Was a full-term child. The presentation was transverse, but the delivery was normal. She is a bottle baby, and soon after birth had a "bad cough," with which she was very ill. She had whooping cough at two months and influenza at five months. She recovered well from all these illnesses and is now well and healthy, except for a small umbilical hernia. The deformity of the head was definitely present at birth.

On examination, she is a plump, rosy-cheeked, healthy-looking child. The head is oxycephalic in shape, resembling rather the traditional helmets of the Roman legionaries. The cranial vault is high, the highest point being situated one inch behind the bregma. Behind this the head slopes gently away for three inches, then descends almost vertically to the external occipital protuberance, which is unobtrusive but can be identified. The parietal eminences appear to be absent, but there is bulging of the temporal regions on both sides, sufficient to hide the ears from view when the head is regarded from the front. There is no sign of either anterior or posterior fontanelle, and no sutures can be felt. The forehead ascends with a slightly forward inclination for 1½", almost to the bregma. Its most prominent part is in the midline, from which it slopes away on either side, the frontal eminences being absent. The superciliary arches are very faintly defined, and above them on each side is a definite grooved depression. The bridge of the nose is broad and flattened to a greater extent than usual at this age, and this gives an appearance of "far-apartness" to the eyes, which are, however, no further apart than normal. The nasal bones are further deformed, the lower portions passing directly forwards at right angles to the upper parts. The face is triangular in shape, though this is partly disguised by the chubby cheeks. The palate is narrow and very highly arched and no teeth are present. The lower jaw is perhaps slightly prominent, though this is relative rather than actual. There is no exophthalmos. There is double internal strabismus but no nystagmus. The pupils are small, equal and concentric; they react normally, and the optic discs are normal. There are no ocular palsies. The following measurements are of interest:—

Greatest circumference, 16".

Nasion to external occipital protuberance, 9".

Bi-parietal, 12".

Bregma to symphysis menti, 7".

There is a slight degree of webbing of the second and third toes of the left foot, which also shows a degree of talipes deformity. The left leg is half-an-inch shorter than the right. Joint movements are free and full in both upper and lower limbs. There is free extension at both elbows, but both give a slight click when full extension is insisted on. Sight and hearing by ordinary methods are normal. The child sits on its mother's knee, bright and lively, reaching out for things and turning her head to a sound, and takes a great interest in her surroundings. She does not stand yet, but the mother states that she has said "Dada" to her father.

X-ray of Skull. (D. F. and M. F.) Both skulls show exactly similar changes (Cases 2 and 3.)

In lateral view the skull appears abnormally thick, and there is no well-defined pituitary fossa. No digital markings are present. On antero-posterior view the head is slightly asymmetrical, and abnormal shadows are seen, due to the enlarged greater wings of the sphenoid. (Dr. B. Leggatt.)

Case I at 9 months.

CASE 3. Mrs. F. Aged 40. Is the mother of the previous case. Her birth was normal as far as she knows, but she was one month premature. She was a breast fed baby and has always been healthy. She worked as a dressmaker before her marriage, and still does fine sewing. She has never suffered from headaches at any time, even after reading, and does not wear glasses now. The deformity of the head was definitely present at birth.

On examination, she is a healthy-looking woman of normal stature. The head, though, of course, on a larger scale and with one or two minor differences, resembles the infant's head with ludicrous exactness. The slope of the head, the temporal bulging and the shape and inclination of the forehead are all faithfully presented. The most obvious difference is one which might be expected, and that is the nose. This is broad across the bridge itself, measuring a quarter of an inch across, but falls vertically to the level of the eyes on either side. The anterior nares are deep and roomy. The only other difference of note is the fact that the face is not markedly narrowed, and the palate is normal in shape. The teeth are absent, except one carious incisor. There is no exophthalmos or nystagmus, while there is a slight double internal strabismus, just as in the case of the child. The optic discs are normal. There are no other abnormalities of any kind, and the special senses are unimpaired. As stated above, she still does fine sewing.

She is quite up to the average in intelligence, and though garrulous is distinctly "all there." She states that when she was a dressmaker she was accustomed to take all her customers' measurements and write them down afterwards.

Family History. Her husband and his family are normal. A maternal cousin, female, has a similar deformity and is mentally deficient (unfortunately we had no opportunity of examining this patient). She has five children, four of whom are in every way normal. The youngest is our second case. There is nothing further of any interest.

In discussing these cases it is proposed to compare them one with another, rather than to regard them separately, and to consider any possible relationship they may have to hypertelorism. The bony abnormalities of the skull itself

Cases II and III.

will first be reviewed, the clinical side of the problem being dealt with later in the light of these.

The work of Greig on these conditions is classical, and those interested in the finer anatomical details are referred to his articles ⁽²⁾ ⁽³⁾.

A consideration of the radiological reports with, in the first case, the post-mortem findings, studied in conjunction with Greig's description of hypertelorism, shews that each of these deformities is associated with a definite developmental abnormality of the sphenoid bone. This sphenoidal abnormality was emphasised by Greig, who shewed also that it begins very early in development.

In the first case, one of scaphocephaly, the radiogram showed considerable under-development of the greater wings of the sphenoid, which was confirmed post-mortem. In the two cases of oxycephaly the greater wings were seen to be grossly enlarged. In hypertelorism, on the other hand, Greig found the lesser wings to be very much larger and more solid than normal, being as large as, or larger than, the greater wings.

In scaphocephaly and oxycephaly the relationship of the sphenoidal abnormality to the deformity is easily seen. In the scaphocephalic skull the poor development of the greater wings of the sphenoid, by partially removing their "scaffolding" function, necessarily brings about a narrowing of the whole skull, especially the anterior part. This narrowing has two secondary effects, namely, the elongation which has been noted, and the degree of elevation of the vault, with absence of frontal and parietal eminences. The generally small size of the orbits, which are not relatively shallow, is also due to the abnormal development of the sphenoid. The greater wing of the sphenoid forms the posterior part of the lateral wall of the orbit, and the under-development of this important structure, with consequent general narrowing of the skull, brings about the generally contracted condition of the orbit found at autopsy. One would not expect to find any relative shallowness of the orbit in this case.

In the cases of oxycephaly the shape of the skull is brought about in the same way, although the difference in the shape of the sphenoid brings about an altogether different deformity. Here the abnormally large solid greater sphenoidal wings push the temporal bones laterally, leading to temporal bulging, with an increase in the transverse diameter of the skull at this point, and wide, shallow orbits. The skull must of necessity adapt itself to this, and the occiput comes forward, cramping the base of the skull, as is seen by the crowding of the foramina in this region, and shortening the antero-posterior diameter. Thus the skull is broader and shorter than normal. The natural consequence of this state of affairs is the great increase of the height of the cranial vault, which has given the condition its name, and the flattening of the occipital region.

Other features of these two types of skull, for instance, the maldevelopment of the maxillæ, which causes the highly arched palate, the thinness of the skull bones with absence of diploë, and the digital markings so frequently met with, shew that other factors are present; but the fundamental fact is the abnormal development of the sphenoid. One further point must be mentioned. Both these deformities are associated with *total absence of sutures*; "where the bones touch they fuse," as Greig has said.

On turning to hypertelorism, the relationship is less evident, and at first sight appears doubtful. The skull is not sutureless, though the sutures are incomplete, and do not form digitations; and a number of intersutural bones are present, internasal, parieto-mastoid and pterionic. It is desired, however, to focus attention on the sphenoid, and consider how the abnormality there affects the problem. The lesser wing of the sphenoid forms the posterior part of the roof of the orbit, and its anterior border articulates

with the orbital plate of the frontal bone. It will be seen that an enlargement of this wing, so gross as in the case in hypertelorism, will push the whole frontal bone upwards, forwards, and outwards, and with it the corresponding maxilla. The nasal bone of that side, which articulates laterally with the frontal process of the maxilla, is naturally involved in this displacement of the entire corresponding side of the face. It is thus easy to understand the necessity for an internasal bone and the retroussé character of the nasal bones themselves. The interfrontal groove noted by Greig is readily accounted for in the same way, while the tilting of the frontal bone which must necessarily occur explains the combination of the low forehead with slight prominence of the frontal eminences.

Cases II and III.

On these grounds is based the contention that the "far-apartness" of the eyes and divergence of the orbital axes, the two most distinctive features of hypertelorism, are directly due to the overdevelopment of the lesser wings of the sphenoid.

This review of the problem, brief as it is, suffices, it is believed, to shew that the origin of these deformities, though they differ in many ways, is the same, namely, abnormal development of the sphenoid bone.

The clinical side of the question will now be considered.

Of associated deformities, symmetrical and otherwise, on which so much stress has been laid, little will be said. This is not because their great importance

is not appreciated, but because the exhaustive treatise of Park and Powers leaves nothing to say. It is merely desired to point out that similar deformities occur in both scaphocephaly and oxycephaly, and in considering the absence of such deformities in cases of hypertelorism so far reported, to refer to a case which was shown to the Royal Society of Medicine by Dr. Robert Hutchison in 1910. This case is referred to by Greig, who gives it as his opinion that, though shown as a case of oxycephaly, it is in reality a case of hypertelorism. On examining the report of the case, and studying the photograph, the case appeared to us to partake of the characters both of oxycephaly and hypertelorism. The head is said to have been oxycephalic, though the photograph gives no confirmation of this, being a "full-face" view. The frontal eminences are definitely absent, and the superciliary arches are ill-defined. There is no temporal bulging to be seen, however, the nose is remarkably broad and flattened, and there is obvious "far-apartness" of the eyes. Other points of resemblance to hypertelorism are the presence of an interfrontal groove, a divergent squint and, what is well shown in the photograph, subcutaneous thickening. In association with the cranial deformity there was syndactyly with "irregularly arranged toes." This is a case which seems to form a link between oxycephaly and hypertelorism, in association with which were deformities of the limbs. The presence of deformities in these cases is, however, inconstant, and it is felt that their absence in hypertelorism does not in any event invalidate the hypothesis proposed.

It is surprising that instances of a familial history in these cases are so uncommon. In explanation of this we quote Greig:—"In oxycephaly as a congenital defect heredity ought to occur, but examples are restricted since those but slightly affected do not seek medical advice and those grossly affected are not acceptable in marriage."

Our second and third cases, however, provide an example of a definitely hereditary abnormality of cranial development. The mother's cousin was similarly affected, and in addition was mentally defective. This is emphasised, as it lifts these conditions out of the "freak" class, and gives them a position of great practical importance.

Slight mental deficiency, which occurs in most cases of malformation of the skull, is merely a natural effect of the very early developmental abnormality of the skull on the growing brain, and is fully discussed by Greig. The other clinical characteristics are all secondary to the cranio-facial deformity or to one another. Little, therefore, can be argued from their occurrence or non-occurrence in a particular case. For instance, the exophthalmos is due to the inadequate accommodation provided by the orbital cavities for the eyeballs in the majority of cases of oxycephaly and scaphocephaly. In Cases II and III there was no exophthalmos. This means simply that the orbits were not as shallow as usual, and therefore the eyeballs could be accommodated comfortably, and by no means invalidates the diagnosis. Nystagmus is common, and

is secondary it is believed to the exophthalmos when present, being produced in the same way as miners' nystagmus. Optic atrophy is another common concomitant of all these conditions, but like exophthalmos depends on the degree of deformity of the orbit and optic foramen.

CONCLUSION.

Scaphocephaly, oxycephaly and hypertelorism are believed to bear a definite relationship to one another, and this relationship consists in a common origin; namely, an abnormal development of the sphenoid bone.

The association of mongolian characteristics with hypertelorism noted by Greig, Cockayne and Braithwaite⁽⁵⁾ appears to be of great importance, though its significance cannot be assessed. The occurrence of epilepsy in a case of Cockayne's⁽⁶⁾ may be mentioned in this connection.

No résumé of the literature has been attempted, but we desire to acknowledge our debt to the following authors, whose work we have studied, in addition to those mentioned in the text:—H. Morley Fletcher⁽⁷⁾ ⁽⁸⁾, R. C. Jewesbury and J. C. Spence⁽⁹⁾, H. C. Cameron⁽¹⁰⁾ and Dr. C. Muir⁽¹¹⁾.

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