THE MORPHOLOGY OF THE SKULL IN A RARE TYPE OF DOUBLE FORMATION

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Double formations ('Doppelbildungen') are defined in Schwalbe's text-book[1] as congenital malformations in which the body axis is always duplicated. There are 2 vertebral columns and 2 central nervous systems. These monstrous formations usually present 2 heads and thus another name for this group in teratology is *dicephalus*. The differences in the various types of dicephalus reside in the degree of duplication of the trunk and of the limbs. The present specimen is a specially rare type of double formation for the reason that the dichotomy was minimal. There was so little separation in the head region that the specimen was considered to be an instance of *diprosopus* or face duplication, until an X-ray examination revealed that the vertebral bodies were double.

The specimen formed part of a collection of teratological material housed in the department. The brain had been removed previously and was not available for study. By kind permission of Prof. L. H. Wells I was allowed to make a dissection of this rare congenital malformation. In view of the fusion in the head region, I concentrated my attention more particularly on the partly duplicated and partly fused skull, the morphology of which presented a fascinating problem.

DESCRIPTION OF THE SPECIMEN

The photographs of the front and back aspects of this prematurely born monstrous infant (Figs. 1 and 2) clearly show the external features. No clinical history was available. The sex was female and the degree of skin pigmentation suggested that the infant belonged to the Cape Coloured population group. Judging by the crown-rump length of 19 cm., the intra-uterine age reached was between $5\frac{1}{2}$ and 6 months.

The face was almost completely duplicated (Fig. 1). The gap between the adjacent angles of the two inner eyes was 20 mm., and that between the adjacent angles of the two mouth openings was 32 mm. The skin stretched smoothly across the median area but a small pit was noted in the 'midline' at about the level of the nostrils, and this depression admitted a probe to a depth of about 2 cm. in an upward direction; the dimple which indicated the opening of the pit is clearly seen in Fig. 1.

Fig. 1. Photograph of the front aspect of a rare double formation.

On the back aspect (Fig. 2) was seen a large spina bifida with total destruction of the lower half of the spinal cord and of the root ganglia. After the remains of the spinal meninges were removed, the stumps of the lower 6 thoracic and of the lumbar and sacral nerves became visible at the level of their respective intervertebral foramina. The partly

Fig. 2. Photograph of the back aspect of a rare double formation.

ossified cartilaginous prominences on each side of the 'neural area' are the remains of the pedicles and transverse processes that survived the destruction of the cartilaginous vertebral arches. For reasons given previously,[2] I attribute such defects to an extensive internal hydrops that affected the lower portion of the neural tube and by fluid pressure caused destruction of the parts. The spina bifida lesion in this specimen must be looked upon as a separate congenital defect in no way related to the partly duplicated head and face.

Dissection. The soft parts were dissected away in order to make a permanent skeletal preparation. Nothing special was noted in the course of removing the abdominal and thoracic viscera. When dissecting the front of the neck from below upwards a gradual transition towards duplication was noted. The single pharynx widened out until there was a large common nasopharynx communicating with the 2 nasal cavities on each side. A common soft palate stretched across the whole area, showing 2 uvulae placed symmetrically on each side. The mandibles had shared in the dichotomy of the facial skeleton and thus there existed 2 mouth cavities and 2 separate tongues, but the latter were fused at their pharyngeal ends.

The skeleton of the trunk. The ossification of the skeleton of the trunk and limbs had proceeded to the stage expected in a normal $5\frac{1}{2}$ - 6 months foetus. The apparently single vertebral column was wider than normal, especially in the lumbar region, and this was at first thought to be a secondary effect from the hydrops distension and destruction of the spinal cord. The spinous processes, still cartilaginous, were single at the back of the neck and upper part of the thorax down to the 5th thoracic. Below T5 the spinous processes, vertebral arches and skin coverings had disappeared to give place to the wide 'neural area', which showed remains of the spinal meninges and the beginnings of the spinal nerves in the intervertebral foramina. The X-ray photograph (Fig. 3) clearly showed that the wide vertebral column was in fact a duplicated one, and that the specimen came into the group of double formations and was not a true diprosopus. The bodies of the vertebrae are duplicated as far down as the 1st sacral, but the rest of the sacrum is single. The bodies of the 9th

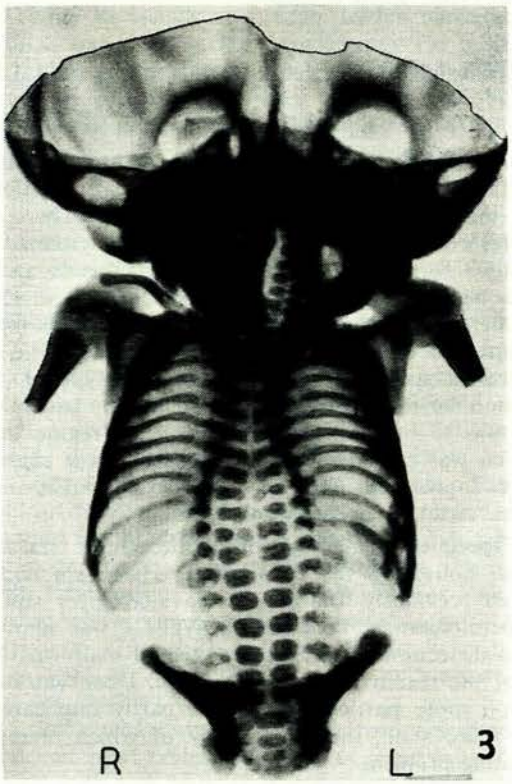

Fig. 3. Radiograph of the skeleton of a rare double formation.

and 10th thoracic vertebrae on the left side, and that of the 11th thoracic on the right side show 2 centres of ossification. This is not unusual. The bodies of the vertebrae occasionally have a pair of centres that coalesce, but may fail to do so.[3] The upper margin of the spina bifida defect can be detected at the upper border of the 6th thoracic vertebra (Fig. 3).

The front aspect of the duplicated skull (Fig. 4). The area between the adjoining frontal bones showed deficient ossification and the gap was bridged by a firm, fibrous

membrane, but in the region between the two 'inner' orbits the greater wings of the sphenoid bones were ossified and were fused across the 'midline'. The two 'inner' zygomatic bones were touching and fused along their margins. A common mass of bone in the 'midline' linked the fused zygomatic bones with the fused greater wings of the

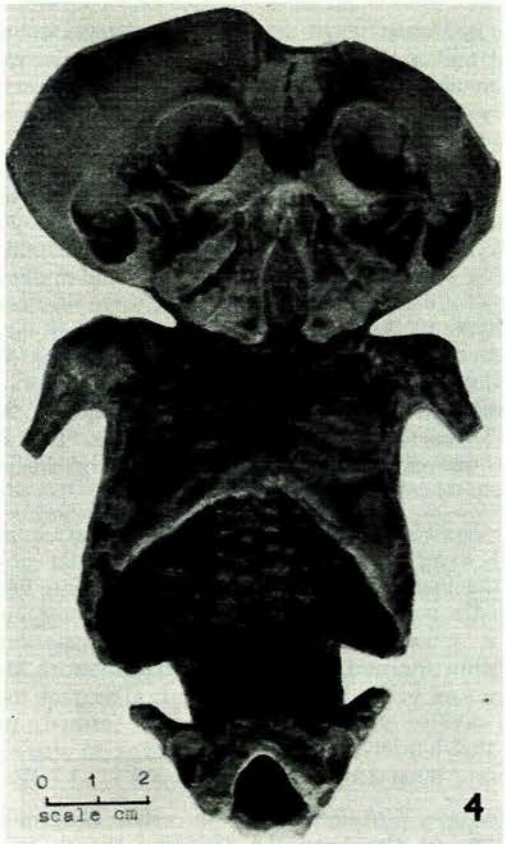

Fig. 4. Photograph of the front aspect of the duplicated skull.

sphenoid, thus forming a kind of 'bridge' across the space. A narrow temporal fossa was found on each side of this 'bridge', leading to the coronoid processes of the duplicated mandibles. In preparing the skull two fused temporal muscles, much reduced in size, were removed. The adjacent heads of the mandibles, however, could not be defined, because they were merged into this common mass of bone that separated the two 'inner' temporal fossae. The skin invagination, described above, ended blindly below the 'bridge' of bone, and no doubt this represented an attempt at duplication of the two external auditory meatus canals that would have appeared if the duplicated faces had become more fully separated. The duplicated orbits, the two anterior bony apertures of the nose, and the duplicated maxillae, were normally formed. The mandibles were separate except for the fusion of the two adjoining heads of the mandibles.

The base of the duplicated skull (Fig. 5). The upper surface of the base demonstrates best the complex morphology of this skull, with duplication of the anterior cranial fossae and sella turcica region and partial duplication of the middle cranial fossae, while there is an absence of duplication of the posterior cranial fossa. The *frontal bones* are completely duplicated, the two interfrontal sutures when prolonged backwards forming a forward directed angle of a little more than 90°. There are 2 complete *ethmoid bones* together with duplication of the other bony elements of the nasal cavities. The *sphenoid bones* are almost fully duplicated. The bodies of the sphenoid bones have swung apart causing the formation of 2 hypophyseal fossae, each bordered behind by a dorsum sellae and presenting a full set of clinoid processes. Further, each pair of lesser wings is duplicated. The only part of the sphenoid that has not undergone complete dichotomy is that pair of greater wings which adjoin the 'midline'. These adjoining greater wings are shortened from side to side and there was bony fusion across the midline, as already mentioned. A radiograph showed that the bodies of the sphenoid bones were separated from the basilar part of the occipital by a plate of cartilage, which was about 2 mm. thick. There was no duplication of the *temporal bones* and the *occipital* bone was a single one. However, the basilar part of the occipital bone was wider from side to side than usual and ended in an angulation with a plate of cartilage on each side, separating the occipital bone from the duplicated bodies of the sphenoid bones. A fold of dura mater is visible, occupying the front part of the 'midline' of this complex cranial cavity (Fig. 5), indicating that there was a partition between the two 'inner' temporal lobes of the duplicated cerebral hemispheres.

DISCUSSION

Double formations are accepted as representing a partial twinning—in other words, a failure to achieve the formation of a pair of homonymous or uniovular twins. Their

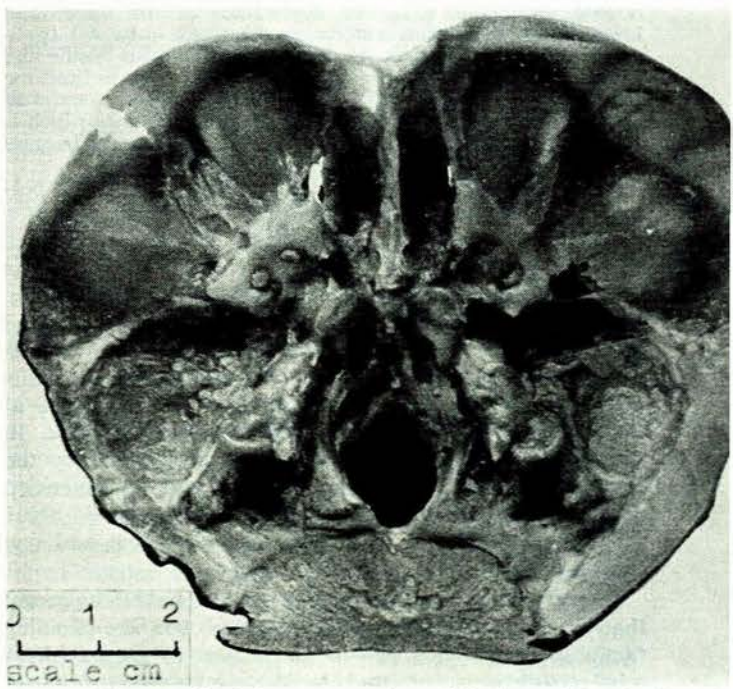

Fig. 5. Photograph of the upper surface of the base of the duplicated

origin must be sought at the embryonic-plate stage. When the split of the embryonic plate affects the cephalic end only, the term used in teratology is *katadidymus*. The monsters are *terata katadidyma*, meaning twinning from the head-end downwards. Such formations are often described as *dicephalus*, adding *di-*, *tri-*, or *tetrabrachius*, according to the number of upper limbs. In all dicephali, however much the trunks appear fused, the body axis is duplicated; there are always 2 vertebral columns and separate central nervous systems.

Similarly, *anadidymus*, which is rarer, means a twinning from the caudal end upwards. These monsters show a splitting beginning in the lower part of the trunk with greater or lesser degrees of duplication of the lower limbs, but the upper part of the trunk may also be involved. In *anakatadidymus* the dichotomy affects both the cephalic and caudal ends of the embryonic plate, the trunks usually remaining fused in the lower thoracic and upper abdominal regions. The fusion may be limited to the skin and superficial tissues, the best-known instance of this being the Siamese twins. A very little further separation would have produced a pair of homonymous twins.

The small collection of teratological deformities in the department contains 7 specimens of double formations. These can be grouped as follows: One, a rare *anadidymus*, with extensive duplication of the trunk and a double set of upper and lower limbs, *tetrabrachius tetrapus*, the heads having remained fused; this specimen can also be referred to as a *syncephalus*. Two instances of *katadidymus*, one a *katadidymus dibrachius*, and the other a *katadidymus tetrabrachius*. The heads are completely separate and such specimens may also be called *dicephalus;* there is no separation in the lower part of the trunk.

The remaining 4 specimens are instances of *anakatadidymus*. One is to be described as *a. tribrachius tripus;* the trunks had not separated sufficiently for the growth of two extra limb buds, the result being a composite 3rd upper and lower limb. The other three are instances of the more ordinary type of double formations with full duplication of the upper and lower limbs, the fusion concerning the trunk only. All specimens of anakatadidymus, of course, have separate heads and could also be called dicephalus. However, it is best to limit the term dicephalus to the true katadidymus type. To this series of double formations is now added the present specimen, which is at the other extreme end from the katadidymus type, because there was a minimum of body-axis duplication.

The degree of dichotomy in the present instance is perhaps the least that is compatible with a classification as a double formation. As far as the body axis was concerned, the only part definitely duplicated was the notochord, as shown by the doubling of the bodies of the vertebrae, including the basilar part of the occipital bone, where the notochord ends. At the caudal end the notochord duplication was not complete; the sacrum from S2 downwards was not split (Fig. 3). The neural tube was duplicated at the forebrain end, but was probably single otherwise. In the spinal-cord area the picture was complicated by the existence of an extensive spina bifida. A hydrops distension and destruction of the parts by fluid pressure would argue in favour of a single cerebrospinal-fluid system and one central canal.

In a previous anatomical study of dicephali[4] I suggested that the splitting of the entoderm layer was the deciding factor in determining whether a monster of the katadidymus, anadidymus or anakatadidymus category resulted. When the entoderm cleavage affects the foregut and varying lengths of the midgut, the result is a katadidymus or dicephalus, with the trunk separation and upper-limb duplication in proportion to the extent of the midgut involved. In the present specimen only the extreme upper end of the foregut became split, viz. the part giving rise to the nasopharynx and oral pharynx. The accompanying neural-tube dichotomy involved only the forebrain.

The forebrain duplication. The forebrain became doubled and a dichotomy of all its derivatives resulted. Thus, on each side there were 2 optic vesicles with the olfactory outgrowths between each pair. There was a median outgrowth from the floor of each forebrain, with duplication of the hypophysis cerebri. The brain capsule showed the adaptive response and the result was the complex skull described above, with 4 orbits, 2 nasal cavities, and a duplicated body of the sphenoid bone presenting a sella turcica on each side of the midline. The brain was not available for study, but there was certainly some degree of duplication of the cerebral hemispheres, as shown by the median partition of the dura (Fig. 5). The frontal lobes and the anterior ends of the temporal lobes were probably double. But the close apposition of the two 'inner' halves of the cerebral hemispheres had almost certainly prevented an effective dichotomy of the parietal and occipital lobes.

The mandible duplication. The mandibles are duplicated (Fig. 4), except for the two adjacent condylar processes where the merging of the parts in the 'median line' had prevented a complete dichotomy. The embryonic interpretation is somewhat difficult, because the pharynx was single below the oral part, and no duplication of Meckel's cartilage was to be expected *a priori*. The fact that two mouth cavities and separate tongues formed, perhaps caused this border region of the pharynx to share in the dichotomy; thus, 2 mandibles appeared.

Comparison with diprosopus. In certain congenital malformations of the head the twinning theory is not a satisfactory explanation of their origin. These malformations are described in text-books of pathology as *diprosopus* —diprosopus diophthalmus, triophthalmus or tetraophthalmus, meaning double-faced with 2 eyes, 3 eyes or 4 eyes, respectively. The body axis shows no duplication. These deformities, according to Schwalbe,[1] are not true double formations but belong to a group which the author calls 'überzählige Bildungen', best translated as excessive formations or growing-end duplications.

Some years ago[5] I described 2 cases of face dichotomy in vertebrates. One, a *diprosopus triophthalmus* in a newly-hatched chicken, the centrally placed 3rd eye being formed by the fusion of 2 adjacent optic vesicles. The other, a *diprosopus diophthalmus* in a calf in which only the nasal cavities were duplicated, without the appearance of a midline eye.

The diprosopus type of malformations are best explained by the so-called growing-point theory,[6] which is based upon our knowledge of the growth of a plant, the tip of the stem being the growing point. The most distal pair of cells divide into two, four, etc. When duplication occurs, the cells behind and between them grow actively, push forwards between the cells at the tip and separate them in

a lateral direction; thus they bring about the appearance of 2 growing tips. In vertebrate embryos one of the growing points at the cephalic end is the forebrain extremity of the neural tube.

The main differences between the specimen of a doubled face which I have described above (Fig. 1) and the specimens of face dichotomy in the 2 vertebrates were as follows: Considered as a true diprosopus, the present specimen would have been labelled a *diprosopus tetraophthalmus* with mandible duplication. But in the chicken and the calf there was no dichotomy of the mandible. In the chicken the lower half of the beak and a single tongue presented midway between the duplicated upper halves of the beak which pointed laterally. In the calf's head the single mandible and tongue were turned upwards onto the face between the doubled nostrils and maxillae. At the base of the skull the present specimen showed a complete duplication of the anterior cranial fossae, which had swung so widely apart that a portion of the duplicated middle cranial fossae appeared on each side of the midline; further, in the central area the hypophyseal fossa was doubled (Fig. 5). In the 2 vertebrate specimens the doubled anterior cranial fossae showed less separation, and no dichotomy of the pituitary fossa could be seen. In all three specimens the crania clearly demonstrated an adaptive response of the brain capsule corresponding to the degree of forebrain duplication that had occurred.

To what extent the hypophyseal-fossa duplication in the present instance can be related to the mandible dichotomy must remain an open question. In the two vertebrate specimens there was no foregut dichotomy and thus no duplication of Meckel's cartilage. Nevertheless, the borderline case of double formation that I have described here raises some doubt whether such a clear-cut distinction can be made between double formations and diprosopus.

SUMMARY

A rare form of cephalic-end duplication that presented a minimum degree of body-axis dichotomy is described. A radiograph showed that the axis duplication was confined to the bodies of the vertebrae. At the head end only the uppermost portion of the foregut had shown cleavage and, as regards the neural tube, the doubling of the parts was limited to the forebrain and its derivatives.

The complex skull, partly duplicated and partly fused, is described, and the dichotomy or otherwise of the various bones of the cranium is analysed.

The 'twinning theory' is accepted as the explanation of the origin of double formations. The various types that result from the incomplete splitting of the embryonic plate are discussed.

Double formations at the head end are contrasted with diprosopus or face duplication. The origin of the latter is best understood by making use of a plant-growth analogy —the so-called 'growing-point theory'. The morphology of the complex cranium described here throws some doubt on the validity of a rigid separation between these two types of congenital malformations.

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