Case Reports


A Monopodal Sireniform Monster with Dermatoglyphic and Cytogenetic Studies

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The earliest known description of a human sireniform foetus, quoted by Kampmeier (1927) dates back to the first century A.D. Many of the early reports were highly imaginative essays like that of Thomas Bartholin whose paper includes illustrations of a 'sirene natans' complete with mammas and webbed fingers and a picture of the monster actually swimming in the sea (Bartholin, 1654).

Such imaginative descriptions will have been influenced by mythical beliefs in sirens and mermaids. The Greek sirens were singing bird-women who lured sailors to their death, as described by Homer (1945: c. 1000 B.C.) in the Odyssey and interpreted in terms of classical religious mythology by Robert Graves (1955). The mermaid or water-nymph type of siren of nautical tradition originates in the mediaeval folk lore of many countries. Baring-Gould (1875) holds that they derive from early sun or moon deities of the Celtic peoples. There are several forms of mermaid legend but nearly all involve a love relationship between a mermaid or merman and a mortal, and they have inspired much fine art and literature.

The deformity is variously known as symphoderma, sirenomelia, sympus, or in its complete form as monopodia or monomelia, or more simply as sireniform or mermaid foetus. Ballantyne (1904) simplified the classification by referring to cases of complete or incomplete fusion of the lower limbs as monopodia or symphoderma, respectively. He recognized that these infants were stillborn or died within a few hours of birth, and suggested that this might be associated with the frequent absence of kidneys. He gave an accurate description of the limb deformities that may occur and noted the major internal pelvic abnormalities that are associated with the limb deformity. A further review was that of Kampmeier (1927) who included another 51 cases and one of his own. Hendry and Kohler (1960) included in another review an exceptional case in which a horseshoe kidney was present but with complete atresia of the urethra.

The more important papers since that time have discussed two aspects of the condition, the association with Potter's syndrome and the possible role of oligohydramnios (Fritzsche, 1955; Bain, Beach, and Flint, 1960; Bearn, 1960) and a possible sex anomaly (Jolly and Lamont, 1958).

Recent reports have included a case associated with multiple malformations. In addition to the usual abnormalities there was a trilocular heart with a high ventricular septal defect, anomalies of the aorta, and also a tracheo-oesophageal fistula (Williams, 1962). The most recent report was that of Mehta, Binsay, and Purandare (1964) who describe an infant with a single ileum, femur, and tibia but no foot.

A case is reported here of an infant with complete fusion of the lower limbs and a rudimentary kidney on whom cytogenetic and dermatoglyphic studies have been undertaken.

Case Report

Clinical Aspects. A woman aged 29 years delivered twins on January 17, 1965. The first twin, a boy, weight 3060 g, was normal and has since progressed well, apart from slight feeding difficulties. The second twin was a stillborn sirenomph monster of weight 950 g. The placenta was said to be single but was not preserved by the midwife, so the zygosity of the twins is uncertain.

On external examination the stillborn foetus was a monopodal sirenomph monster of total length 35 cm and crown-rump length 23 cm. There was a typical Potter facies and rather large spade-like hands (Fig. 1). The lower extremity had a posteriorly facing knee-joint and two distal joints terminating in a single digit complete with a nail. There was no anus or external genitalia.
but the sacrum formed a small posterior prominence. Radiographs showed a single femur and tibia (Fig. 2), distal to which there was one bone with a small ossification centre. In addition, there were 13 ribs, a hemi-vertebra in the lumbar region (Fig. 3), and a marked thoracolumbar scoliosis with the concavity to the left.

There was gross hypoplasia of the lungs which weighed only 2.8 g. together, histology showing congestion and persistence of cuboidal alveolar epithelium in places. The heart, which weighed 4.5 g., had a hypoplastic left atrium, ventricle, and aortic arch, with pulmonary veins draining into the right atrium and the pulmonary trunk continuing through the ductus arteriosus as the descending aorta. There was a single umbilical artery arising as a midline continuation of the aorta and the lower limb gained arterial supply through a vessel derived from the posterior wall of the aorta.

The oesophagus and stomach were normal. The intestine was unrotated with a blind end and an additional loop of intestine ending blindly at both ends was attached to the left of the main gut by a short mesentery. The liver was extremely small, weighing only 22.5 g. with rather pointed lobes, but appeared normal on section. Histology showed active erythropoiesis. The kidneys, ureters, and bladder were apparently absent, but a small round pink structure of 0.5 cm. diameter on the posterior wall of the pelvis proved on histological examination to be renal tissue. There was a well-defined cortex and medulla, many of the glomeruli appeared immature, and the tubules and collecting ducts were poorly formed with a few microcysts. There were two normal testes within the abdominal cavity. Spleen, pancreas, thyroid, and thymus were unremark-
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Fig. 4. Drawing of dermal ridge pattern of tip of lower limb digit.

Fig. 5. Cell with analysis of chromosomes.
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able. The adrenals, discoid in form, weighed only 1 g together but were histologically normal. Both ilia and pubic bones of the pelvis were present with a single central acetabulum. The range of movement of the lower limb suggested that it was developmentally a left leg, and the position of the nutrient foramen of the femoral shaft confirmed this. The lower end of the femur, however, was unduly broad with two patellae. The lower femoral epiphysis was present but the upper tibial epiphysis had not appeared. The brain weighed 250 g. with a mature gyral convolutional pattern and early myelination within the internal capsule on section.

Dermatoglyphic Study. The dermal ridge patterns of the hands showed no unusual features. On direct inspection, the right hand showed whorls on the tips of all five fingers, a 3rd interdigital pattern, a single axial triradius in the usual t position, and no thenar pattern. Inspection of the left hand showed whorls on digits 1 to 4 and an ulnar loop on the 5th finger, a 3rd and a 4th interdigital pattern, a similar single axial triradius to that on the right, and again no thenar pattern.

Of greater interest was the pattern on the lower extremity where the ridges were poorly formed. They covered the ventral surface of the tip of the extremity, running in horizontal circles proximally, but in longitudinal lines distally with a single central triradius at the junction of the circular and longitudinal ridges (Fig. 4).

Cytogenetic Study. Fibroblast cultures were established from tissue taken at the time of necropsy. Slides were prepared for cytological study according to a modification of the method of Harneden (1960). The chromosomes were counted in a total of 43 cells showing well-spread metaphase plates. Of these, 41 cells contained 46 chromosomes and 2 contained 45 chromosomes. All cells showed 5 small acrocentric chromosomes, and 7 cells analysed in detail from photographs showed the clearly distinguishable Y chromosomes of a normal male karyotype (Fig. 5). Interphase cells were counted for sex chromatin and no Barr bodies were seen, that is to say the infant was chromatin negative.

Discussion

Sympodia is a rare congenital anomaly. Only three cases were found among 7117 perinatal deaths in the Perinatal Mortality Survey (Butler and Bonham, 1963). Assuming a perinatal mortality rate of 4% this gives an incidence of 1 in every 60,000 live births. Despite the rarity of the condition, over 200 cases have been reported including one other instance of an affected male infant with a normal male twin (Hendry and Kohler, 1956).

The clinical and pathological findings in this child show that it had the less common form of the condition, complete fusion of the lower limb, monomelia or monopodia. The internal rotation of the lower limb bud that normally occurs during early development had, as with all cases of sympodia, been prevented by the fusion into a single limb, with the result that the limb faced in a reverse direction to normal. The case is atypical in that a rudimentary midline kidney was present. A similar small rudimentary renal mass was described as lying on the left side of the abdomen in the case reported by Bearn (1960). Hendry and Kohler (1956) report the presence of an inverted horseshoe kidney in one of their three cases.

The child reported here also clearly shows the syndrome of large, low-set ears, flattened nose, epicanthus, receding chin, and pulmonary hypoplasia described by Edith Potter (1946a, b) in infants with renal agenesis. Bain et al. (1960) and Bearn (1960) have drawn attention to the frequency of Potter's syndrome in sympodia, and have discussed the role of oligohydramnios in the syndrome. Both Bearn and Bain and his colleagues regard oligohydramnios as secondary to renal agenesis and as possibly producing some of the facial features of Potter's syndrome. They are agreed that sympodia itself cannot be due to oligohydramnios.

All reviews of sympodia have found an excess of male cases. Kampmeier (1927) found 38 male to 14 female cases. In one case described by Jolly and Lamont (1958) there were primitive testes but female sex chromatin was found in tissue from several sites. This prompted a study of sex chromatin and chromosomes in the present case. The finding of normal testes, normal male karyotype, and absence of sex chromatin means that, in this case at least, the primary defect does not involve sex differentiation. Passarge and Sutherland (1965) in three cases of Potter's syndrome with renal agenesis but not sympodia also found a normal chromosomal constitution.

Wolff (1936, 1948) produced a range of cloacal anomalies, especially anorectal, and sympodial deformities in chicks by irradiating the caudal end of the embryo. The degree of malformation varied with the extent of the tissue damage produced. In man, twins have been reported, one with sympodia and the other with anal atresia and absence of one kidney (Bauereisen, 1905). Bain and Scott (1960) found gross defects of the caudal end of the foetus, such as spina bifida or sirenomelia, in 7 out of 50 cases of renal agenesis. These reports have led to the concept of a variety of congenital anomalies affecting the caudal end of the foetus that may occur in association with one another, including anal imperforation and intestinal anomalies, urinary tract dysplasia, anomalies of the lower end of the spinal column, and sympodia. Duhamel (1961) has termed these anomalies 'the syndrome of caudal regression'.

As to the cause of this spectrum of malformation
there is very little positive evidence. Whether it is due to a failure of development of caudal segments of the embryo, as suggested by Bolk (1899), or to subsequent damage to the caudal segments, is not known. No specific environmental factor has been identified. On the other hand, evidence for a genetic factor is sparse. Only one instance of symphodia in both of identical twins has been recorded (Roberge, 1963), and there are no other reports of familial cases, but nor are there any reports of identical twins with only one affected. An analogous syndrome has been described in the mouse, differing from human cases only in that all the affected mice were female (Gluecksohn-Schoenheimer and Dunn, 1945). Five such mice were observed, arising as isolated cases from five different matings, but each affected zygote carrying some combination of the harmful genes T (short tailed), r^2, or 'u' (recessive tail mutations, the latter also affecting the genito-urinary system) and Fu (fused). These observations suggest that symphodia is not due to a single abnormal gene but is produced by possibly one or more of several environmental factors acting on a predisposing genetic background.

The presence of dermal ridges, with an unusual pattern, on the single digit of this case confirms that the defect originates at an early embryological stage, the ridges being laid down at about the 6th—8th week. The fact that the pattern shows a single triradius without any loops appears to conflict with the rule, proposed by Penrose (1965), that the number of triradii on the palm plus 1 is equal to the number of loops plus the number of digits. However, Penrose showed that the nail might mask a loop of the ventral pattern, and this is probably the case here. The very simple pattern suggests that the volar pad on the ventral surface of the terminal phalanx that normally develops on the foetal digits was not formed here.

Summary

A case is reported of a monopodal sireniform stillborn male foetus. The child was one of twins, the twin being a normal liveborn male. An interesting feature was the presence of a single rudimentary midline kidney. The completely fused lower limb ended in a single digit having an unusual pattern of dermal ridges. Sex chromatin and chromosomal constitution were of the expected male pattern. The features and possible aetiology of symphodia and monopodia are discussed with a brief introductory diversion into the literature of mythical sirens and mermaids.

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References


Addendum

After this report was submitted for publication Johnson (1966) described a further case in which the single lower limb contained two complete lower limb skeletons. He discussed the antenatal complications.

Reference