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Unilateral Potter syndrome with amelia

by

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In a newborn infant, absence of kidney, hypoplasia of the lung, absence of the subclavian artery, of the adnexa, of the upper limb and its acetabulum were found on the right side, together with renal dysgenesis and other anomalies on the left side. It is presumed that the severe anomalies damaging the right part of the body were consequences of an increased intrauterine pressure due to oligohydramnion.

Amelia occurs as a consequence of intrauterine amputation or of mechanical trauma. In primary amelia rudimentary parts of the hand such as fingers or nails frequently occur on the end of the missing limb. In our case the right upper extremity and the glenoid cavity were missing and renal agenesis and other malformations were observed, localized mainly to the right side.

Report of a case

A one day old female infant was admitted with severe cyanosis and total right upper limb amelia. The mother before or during her pregnancy had never received any kind of treatment which could have been in connection with the malformation.

Delivery had been started artificially because of meconium in the amniotic fluid; it was uneventful, the baby weighed 2600 g. The amniotic fluid was normal in amount. The placenta and the amnion were normal. The baby had hypertelorism, a flat nose, micrognathia and low-set ears. The chest was asymmetric and its right part was compressed (Fig. 1). In the right parasternal line a systolic murmur was heard, pointing to a heart defect.

The chest X-rays revealed a shadow running from the right apex in the direction of the diaphragm. The central shadow was displaced to the right. The right scapula was rudimentary. The clitoris seemed to be enlarged. Urine analysis was negative.

Oxygen inhalation was followed by a transitory improvement until on the 10th day a bronchopneumonia had developed on the right side and 24 hours later the baby died with circulatory and respiratory failure.

Chromosome examination of the bone-marrow showed 46XX/45X mosaicism; of the 30 cells analysed, 6

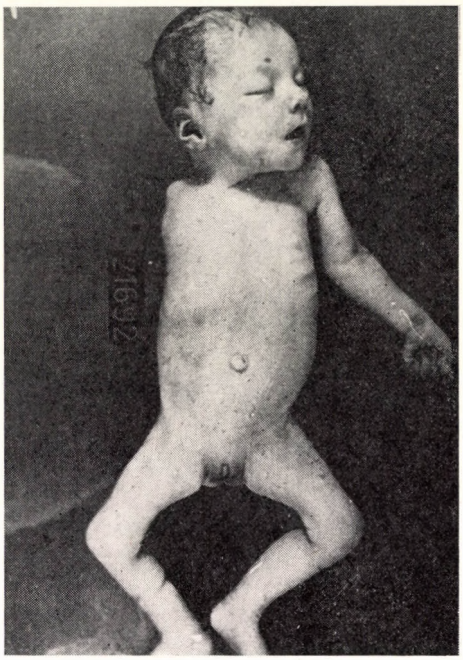

FIG. 1. Amelia with compressed thorax on right side

showed monosomy. Autopsy revealed a total absence of the right upper extremity and of the glenoid cavity. The right scapula and the right half of the thorax were deformed (Fig. 1). The abdominal organs presented an irregular position, with the liver in the centre and its two lobes having the same size. The unicornous uterus was 8 mm long, of pencil thickness on the left side. It had a normal tube and ovary, but on the right side the adnexae were missing. Next to the vertebral column in the height of the right ilium a streak gonad of lentil size was seen, from which a thread-like structure ran in the direction of the inguinal area. The heart was enlarged, in transverse position, dislocated to the right, with the apex directed to the left. Both ventricles were dilated, only the left pulmonary orifice was in the left atrium. while the right orifice was in the slightly dilated right atrium. The foramen ovale was open. The large vessels originated regularly from the aortic arch, but on the right side only the carotid could be detected, the subclavian artery was absent. On the left side, under the origin of the subclavian artery, the dilated ascending aorta narrowed down. Further down its width corresponded to age.

The left lung was normal in size and location, but possessed no lobules. The right lung was rudimentary; in volume it amounted to one third of the left one; upwards it was cone-

shaped and consisted of a unique inferior lobe (Fig. 2). The vessels and the bronchi of the rudimentary right lung were narrow.

The right kidney was absent, the left renal artery somewhat dilated, the left kidney enlarged and the renal pelvis greatly dilated. In the cortical substance of the left kidney small cysts were found. Near to its origin the ureter was constricted (Fig. 3). The left ureter entered the bladder at

FIG. 2. Unilobar lung, considerably smaller on right side

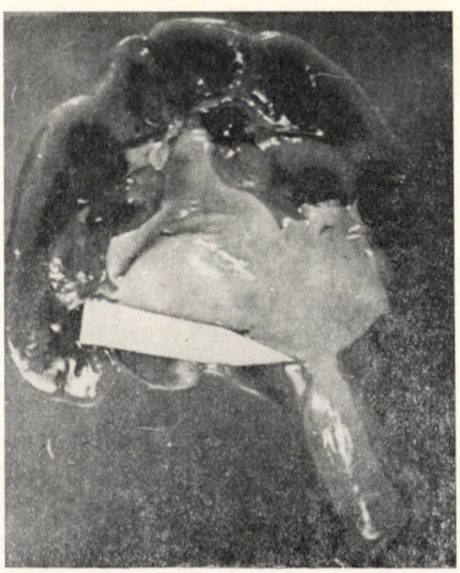

FIG. 3. Constriction on pyelo-ureteral border

the usual place; because of the absence of the right ureter, the trigone was deformed.

In the gonads, identifiable as ovaries, no testicular substance was found. The slightly enlarged clitoris showed a regular structure.

Discussion

In connection with renal agenesis, Potter [7] described a syndrome with a typical face, pulmonary hypoplasia and limb anomalies as the charasteristic symptoms. Since then, in connection with renal agenesis and dysgenesis, various anomalies have been observed [2, 8], and some authors even reported on chromosomal disorders [3, 4]. Still, neither the facial symptoms nor the pulmonary hypoplasia are compulsorily connected with renal agenesis [1]. In the opinion of some authors, the syndrome is due to oligohydramnios, a consequence of the decreased urine secretion. Especially the developmental anomalies of the lungs might be explained by oligohydramnios. According to Maurer et al. [6] even in cases when at delivery no oligohydramnios is present, it may be supposed that in the critical phase of embryogenesis the inhibition of pulmonary development was the consequence of an oligohydramnios.

The role of oligohydramnios in the development of the Potter syndrome was verified experimentally [9]. On the basis of other observations it may be presumed that the pulmonary hypoplasia is the consequence of other anomalies in the thorax such as a decreased thoracic volume due to diaphragmatic hernia, pneumothorax or pneumomediastinum, or leakage of amniotic fluid [5, 10].

Our case differs from the usual ones as in the paired organs the changes were observed unilaterally: absence of the right subclavian artery, rudimentary lung on the right side, presumable absence of the right upper lobe, absence of the kidney on the right side, absence of the right adnexa with streak gonad, absence of the right upper limb and the acetabulum, further a compression of the right half of the thorax. The two last symptoms might be due to an increased pressure affecting the right upper part of the body in intrauterine life.

Because of the renal dysplasia on the left side (ureteral constriction, dilated renal pelvis) urine secretion may have been inhibited also on the left side. It is not easy to answer the question, how far the XX/X mosaicism had influenced the clinical picture. The coarctation of the aorta and the left-side renal disorder are characteristic of Turner syndrome, too.

Unilateral absence of a kidney might involve numerous developmental anomalies [2], but in our case it is presumed that the severe anomalies damaging the right part of the body were consequences of an increased pressure in intrauterine life. This might explain the unusual form of the Potter syndrome.

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