HYDROMETROCOLPOS ASSOCIATED WITH MICROPHTHALMOS, GENU RECURVATUM AND SYNDACTYLY

by

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Hydrometrocolpos is a condition in which the uterus and vagina are distended by retained fluid other than blood. Spencer and Levy (1962) reviewed the literature since 1900 and described details of 62 cases of this abnormality, including 3 cases of their own. Subsequent literature up to 1972 revealed 13 more cases, of which 2 were reported from India (Kawathekar, 1971). The spectrum of associated anomalies in the reported cases included imperforate anus, atresia of the oesophagus and duodenum, bicornuate and double uteri, double vagina, bifid clitoris, congenital urethral membrane, double, stenotic and hydro-, ureters, polycystic kidneys, congenital dislocation of hip, polydactyly, sacrococcygeal teratoma, and hemihypertrophy. The present case of hydrometrocolpos deserves documentation on account of its significant association with microphthalmos, genu recurvatum and syndactyly.

CASE REPORT

A full term female foetus with distended abdomen, small eyes, genu recurvatum and hypertrophied vulva, was stillborn with breech presentation, to a healthy second gravida of 28 years age. The first female child was the result of a 7 months' premature delivery, who died within 2 hours of her birth. Obstetric and family histories were negative and the parents, nonconsanguineous.

The specimen weighed 1520 g and had a sitting height of 33 cm. Placenta and umbilical cord were normal. External examination revealed missing eyebrows, rudimentary eyelashes, and a deeply set and narrow palpebral fissure measuring 7 mm transversely. Eyeballs could not be seen. The nasal bridge was depressed and was crossed by a horizontal sulcus continuous with the line of apposition of the lids on each side (Fig. 1, insert). The circumference of the distended abdomen measured 37 cm. The anus was patent, and meconeum could be squeezed out of it without affecting in any way the distension of the abdomen. The labia majora were large and hanging, and the vaginal orifice large and patulous. No bulging could be seen through the vagina. Subsequent dissection revealed that the vagina ended blindly into a 1.2 cm long atretic segment, although urethra opened into it anteriorly. This was known by the passage of a blunt probe into the urinary bladder. There was bilateral genu recurvatum, and the right third and fourth toes were syndactylous.

Upon dissection, a greatly distended cystic mass arising from the pelvis was noted. Initially a separate urinary bladder could not be identified, and the rectum also was very narrow and adherent to the lower part of the cystic mass. The summit of the cyst demonstrated a partly demarcated, thick walled and cystic uterus measuring 2.5 cm transversely. Arising from each side of the uterus were the uterine tube, round ligament and ligament of the ovary, all firmly adherent to the side of the cystic swelling. The umbilical arteries, uterus and ovarian vessels coursed on either side of the swelling at right angles to the forementioned structures. The cystic mass, along with the urinary bladder and rectum (Fig. 1) was dissected and removed. The mass measured 11.2 cm vertically, and 8.7 cm transversely. The contained fluid, measuring 323 ml, was chiefly serous and partly mucus, mixed with white flakes of desquamated epithelium, and was straw coloured. The urinary bladder was compressed anteroposteriorly and so was the rectum. Both of them were firmly adherent to the lower part of the cyst, but had independent outlets. Longitudinal section of the cyst (Fig. 2) demonstrated clearly a large

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hydrocolpos and a small hydrometros. The cervix was merged imperceptibly with the body of the uterus, which was 1.2 cm thick. The anterior wall of the uterus was 2.6 cm long and the posterior wall, 2.8 cm long; the cavity measured 0.5 cm anteroposteriorly. The openings of the Fallopian tubes within the uterus were not identifiable.

Dissection of the orbits revealed extremely small eyeballs (Fig. 1), each measuring 4 mm transversely and 5 mm anteroposteriorly. Optic nerves were correspondingly very thin. Other contents of the orbit, including muscles and nerves, however, were surprisingly better developed.

Rest of the organs, including liver, spleen, digestive tract, kidneys, ureters, suprarenals, heart, and lungs, were normal, except for dilatation of distal half of the colon.

Discussion

The present case is an example of hydrometrocolpos resulting from atresia of the lower vagina combined with an oversecretion of the foetal uterine and cervical glands consequent to their stimulation by maternal oestrogens. The swollen vulva is a known response to the maternal hormones. Presence of a blind vagina at the vestibule, with opening of the urethra within it, appears to be a result of upward retraction of the vaginal orifice into the pelvis, brought about by the enlarging upper vagina which escaped from the small pelvis into the more roomy abdominal cavity above. When such retraction fails to take place the condition presents as a bulging membrane protruding from the labia, with unusual prominance of the vulva and perineum.

McKusick and his coworkers (1964) have shown that at least one form of hydrometrocolpos is inherited as a simple autosomal recessive. The genetic basis of the anomaly was also supported by Dungy *et al* (1971). Association of the microphthalmos in the present case is unique and significant. Microphthalmos is a condition which either can be inherited as a Mendelian dominant or recessive, or even can be a feature of 13-15 trisomic syndrome (Morison, 1970). Its association with hydrometrocolpos is suggestive of both possibilities: one, that the two traits might be genetically linked, and two, that both of them may be mere phenocopies. The concomitant association of genu recurvatum and syndactyly would favour an environmental causation, i.e. the concept of phenocopy, rather than a genetic aetiology.

Summary

Hydrometrocolpos associated with microphthalmos, genu recurvatum and unilateral syndactyly of third and fourth toes is described in a full term stillborn foetus. The lower part of the vagina was atretic along a length of 1.2 cm. Though there is evidence for a genetic basis of the hydrometrocolpos and that of the microphthalmos in the reported cases, the concomitant association of genu recurvatum and syndactyly in the present case favours an environmental causation rather than a genetic aetiology.

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See Figs. on Art Paper V