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Hydrometrocolpos – A Lower Mesodermal Defects Sequence

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Hydrometrocolpos

Hydrocolpos is the distension of the vagina with sterile fluid caused by vaginal outlet obstruction. When there is associated distension of the uterus it is known as hydrometrocolpos. It is an uncommon congenital disorder with an incidence of approximately 1 in 16,000 female births [1]. Vaginal obstruction may result from vaginal atresia, a vaginal septum, imperforate hymen, persistent urogenital sinus or a cloacal anomaly, depending on the timing and nature of the embryological process that is disrupted [2].

The mass in hydrometrocolpos arises secondary to vaginal outlet obstruction which results in retained vaginal and cervical secretions, which are secreted in response to circulating maternal estrogens [2]. Also, associated urinary and intestinal fistulae are usually present leading to urine and meconium being retained [3].

Hydrometrocolpos can have a varied presentation. It can be diagnosed in utero or it can present at birth or at puberty. Prenatal diagnosis of hydrometrocolpos can be difficult as a result of its rarity, variable presentation and poor ultrasonography. The earliest gestation at diagnosis is reported as 25 weeks and most cases are diagnosed in the third trimester [4]. It can present in the neonatal period as an abdominal mass. At puberty, when hydrometrocolpos is usually secondary to an imperforate hymen, are presents as a bulge with a bluish discolouration at the vaginal introitus [5].

The prenatal differential diagnosis of hydrometrocolpos includes renal lesions, enteric cysts, ovarian cysts, mesenteric cysts, dilated ureter, loops of bowel, anterior sacral meningocoele, pelvic component of sacrococcygeal teratoma and retroperitoneal cystic lymphatic malformations [6].

Hydrometrocolpos can be isolated or associated with other abnormalities. It can be a part of several syndromes. In McKusick-Kaufman syndrome, a rare autosomal recessive disorder, congenital hydometrocolpos is associated with polydactyly and congenital heart disease [7]. Bardet-Biedl syndrome is an autosomal recessive disorder characterised by hydrometrocolpos, postaxial polydactyly, retinal dystrophy or retinitis pigmentosa, obesity, nephropathy and mental retardation. While McKusick-Kaufman syndrome can be diagnosed in very young children, the diagnosis of Bardet-Biedl syndrome is generally delayed upto teenage years. In Ellis-van Creveld syndrome there are polydactyly, acromelia and cardiac anomalies in addition to hydrometrocolpos.

The abnormalities associated with hydrometrocolpos can be primary or secondary [8]. Primary defects arise due to developmental defects whereas secondary effects are due to pressure from the mass. The secondary effects could be hydronephrosis, pulmonary hypoplasia, intestinal obstruction, respiratory distress and leg swelling. The prognosis for hydrometrocolpos is excellent if it is an isolated abnormality such as an imperforate hymen. The prognosis is worse, with significant morbidity and mortality, when there are associated congenital abnormalities which are present in 17 – 81 % of cases [9].

In our analysis of four autopsy cases of hydrometrocolpos at our centre [10] (Figure 1), we identified a spectrum of abnormalities associated with hydrometrocolpos, both primary and secondary. In all four cases there was a significant association of abnormalities. Most notable of these was the presence of pulmonary hypoplasia in all four cases. The kidneys were abnormal in three of the four cases, two were due to bilateral hydronephrosis and the third had unilateral renal agenesis. The external genitalia were ambiguous in three cases. The anus was imperforate in three cases with a high anorectal anomaly in all three cases and a colovaginal fistula in one. The bladder was distended in all four cases with urethral atresia in one case. There were vertebral/skeletal abnormalities in all four cases with sacral dysgenesis in two cases, sacral hemivertebra in one case and bilateral arachnodactyly in the fourth. This non-random association of abnormalities of the genital tract, renal/urinary system, lower gastrointestinal tract and axial skeleton is suggestive of lower mesodermal defects sequence [11].

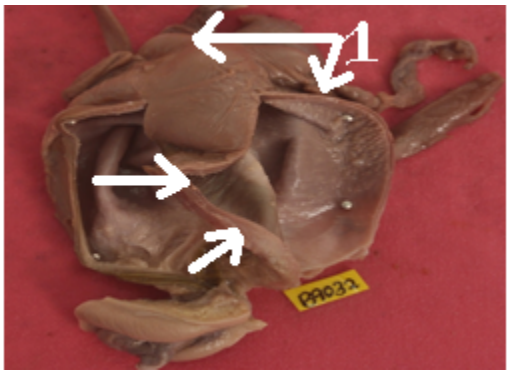

Figure 1: Autopsy cut specimen of distended vagina of a 28 week old fetus showing longitudinal (thin arrow) and transverse (thick arrow) vaginal septa which obstructed the vagina and caused hydrometrocolpos. 1 – Uterus didelphys showing the two horns of the uterus.

Lower mesodermal defects sequence, as described by Pauli et al in 1994 involves the recurrent clustering of abnormalities of the renal, urinary, genital, lower gastrointestinal and axial skeletal systems. All these structures derive from the lower, infraumbilical, portion of the intraembryonic mesoderm. Not all abnormalities need to be present in all cases. But all structures involved share a common embryologic origin. The importance of ascertaining the diagnosis is in helping to counsel the bereaving parents. Firstly, it is the result of a single abnormality, namely, abnormality in the development of the lower

embryonic mesoderm rather than there being multiple 'abnormalities' in the fetus. Secondly, no specific environmental insult has been identified, which means that the condition is not due to something the parents might have done or not done. Lastly, the diagnosis implies little or no risk of recurrence, which helps in reassuring the parents.

References

1. Ayaz UY, Dilli A, Api A (2011) Ultrasonographic diagnosis of congenital hydrometrocolpos in prenatal and newborn period: a case report. Med Ultrason 13: 234-236.
2. Nunes E, Branco M, Meyer F (2014) Prenatal diagnosis of fetal hydrometrocolpos. Acta Obstet Ginecol Port 8: 405-407.
3. Troyano-Luque JM, Alvarez de la Rosa M, Padilla AI, Laura ces, Martinez JM, et al. (2009) Pre- and postnatal features of hydrometrocolpos in one fetus of a dizygotic twin pregnancy. DSJUOG 3: 97-101.
4. Cerrah Celayir A, Kurt G, Sahin C, Cici I (2013) Spectrum of etiologies causing hydrometrocolpos. J Neonatal Surg 2: 5.
5. Vitale V, Cigliano B, Vallone G (2013) Imperforate hymen causing congenital hydrometrocolpos. J Ultrasound 16: 37-39.
6. Taori K, Krishnan V, Sharbidre KG, Andhare A, Kulkarni BR, et al. (2010) Prenatal sonographic diagnosis of fetal persistent urogenital sinus with congenital hydrocolpos. Ultrasound Obstet Gynecol 36: 641-643.
7. Yewalkar SP, Yadav VK, Khadse G (2013) The McKusick-Kaufman hydrometrocolpos-polydactyly syndrome: A rare case report. Indian J Radiol Imaging 23: 183-185.
8. Geipel A, Berg C, Germer U, Ahrens P, Gloeckner-Hofmann K, et al. (2001) Diagnostic and therapeutic problems in a case of prenatally detected fetal hydrocolpos. Ultrasound Obstet Gynecol 18: 169-172.
9. Saxena R, Divan G, Long AM, Arulambalam KJ, Nicolaides KH (1993) Fetal hydrometrocolpos. Ultrasound Obstet Gynecol 3: 360-361.
10. Bodduluri VL, Srinivasan L (2015) Hydrometrocolpos - A lower mesodermal defects sequence. J Fetal Med 2: 191-196.
11. Pauli RM (1994) Lower mesodermal defects: a common cause of fetal and early neonatal death. Am J Med Genet 50: 154-172.