Imperforate hymen in a sixteen-month-old child

The hymen is a solid membrane located at the entrance into the vagina. The most common form is the annular hymen with one central opening. However, there are several other variations such as the microperforate hymen, hymen with multiple perforations, septate hymen etc. If, during development, the central opening does not form on the hymen for any reason, this type of hymen is called the imperforate hymen. The imperforate hymen is a congenital abnormality in the development of the female reproductive system which, if it remains unidentified by that time, is mainly diagnosed in the age of puberty, i.e., with the onset of the first menstrual bleeding. In that period, the clinical picture varies from abdominal pain, lower back pain to acute urinary infection. Once a diagnosis is made, a small surgery, hymenectomy, is performed in order to create the central opening of the hymen. From what has been said, one could infer that this is not a serious anomaly, which is somewhat true if we talk about an imperforate hymen diagnosed with the onset of the first menstrual bleeding. However, the diagnosis of an imperforate hymen in a young child, as is the case with the patient whom we present in our paper, requires a thorough examination due to possible presence of other anomalies of the genitourinary system. The reproductive and the genitourinary systems are closely connected during their embryonic development. In the sixth week of embryonic development, both male and female embryos have two sets of paired ducts, the so-called Müllerian ducts, from which parts of the female reproductive system such as the uterine tubes, uterus, cervix and the upper two thirds of the vagina develop, as well as Wolff canals from which parts of the male reproductive system (vas deferens) develop. The lower part of the vagina develops from the urogenital sinus, whereas ovaries develop from germinative cells. The disruption of this development leads to the occurrence of various anomalies, of which one of the most serious is the absence of the uterus – the anomaly known as the Mayer-Rokitansky-Küster-Houser (MRKH) syndrome. This syndrome, despite the otherwise normal female karyotype – 46XX, is characterised by the absence of the entire uterus, the upper two thirds of the vagina and both tubes. External genitalia develop normally. The ovaries are normally developed and their function is preserved. This state is also known as MRKH syndrome type I, which is usually less common, unlike the MRKH syndrome type II, which is unfortunately more common, and which is usually accompanied by the associated kidney, spine, hearing apparatus and heart anomalies. This is why we lay emphasis on a thorough examination of external genitalia in female children, which is also the subject of our paper “Imperforate hymen presenting as vaginal cyst in a 16-month-old child – considerations for an early diagnosis” published in the *Scottish Medical Journal.* Another possible outcome of differential diagnosis which is very similar to an imperforate hymen in newborns and small children is the Skene’s duct cyst. These are the ducts that are considered to be homologues of the male prostate, and since they are located near the urethra, a diagnosis is usually made based on the location of the cyst relative to the outer opening of the urethra and vaginal patency. Given that a cyst can dislocate the mouth of the urethra with its location and size, this state, unlike the imperforate hymen in small children, requires a surgical intervention.

The symptoms of anomalies in the development of the Müllerian ducts depend on the patient’s age. In newborns and small children, a palpable mass in the abdomen, pelvis or vagina may be

considered as the first sign, whereas in adolescents, the first menstrual period is usually delayed. Adult women, on the other hand, most often turn to doctors due to infertility.

The cause of these anomalies is not yet fully known. Nonsteroidal estrogen, which is often recommended to women with high-risk pregnancies, is mentioned as one of the possible external causes. Today, however, the thesis of the genetic cause of the anomaly receives a growing support. The gene that the research so far most associate with the forming of the female reproductive system is the WNT4 gene. It has a proven role in the regulation of cell and tissue growth and differentiation during early prenatal development.

In our patient, a girl aged 16 months, the first symptom noticed by her mother was a smaller swelling at the entrance to the vagina accompanied by baby's cries. Such a condition, considering her age, required a thorough examination which resulted in the finding of the imperforate hymen.

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