CASE REPORT

Familial Imperforate Hymen

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Abstract

Imperforate hymen, a rare obstructive anomaly of female genital tract, is accepted to be sporadic and isolated. We report an unusual case of familial imperforate hymen and discuss the possible inheritance mode. This is the fifth case to be reported. In our case, there were two affected sisters in a consanguineous marriage suggesting recessive mode of transmission. Imperforate hymen could be a familial condition and examination of family members is important to avoid late diagnosis and associated complications.

Keywords: familial imperforate hymen, inheritance, diagnosis

Özet

Ailesel İmperfore Himen

Kadın genital sisteminin nadir bir obstrüktif anomalisi olan imperfore himen genellikle sporadik ve izole olarak kabul edilir. Bu yazıda, nadir bir ailesel imperfore himen vakası bildirilmiş ve muhtemel kalıtım yolları tartışılmıştır. Literatürde bildirilen beşinci vaka olan bu raporda, akraba evliliğinden doğan etkilenmiş iki kız kardeş olası bir resesif kalıtımı düşündürmekteydi. İmperfore himen ailesel bir durum olabilir. Geç tanıya bağlı komplikasyonların önlenmesi için aile bireylerinin de muayeneyle taranması gereklidir.

Anahtar sözcükler: ailesel imperfore himen, kalıtım, tanı

Introduction

Imperforate hymen, occurring in 0.1% of newborn females, is a rare obstructive anomaly of genital tract. In this uncommon condition, canalization of hymen fails causing hematocolpos and hematometra after puberty. Imperforate hymen is accepted to be sporadic and isolated. Therefore, family members are not screened for imperforate hymen in general practice. However, we report an unusual case of familial imperforate hymen.

Case

A 16-year-old, unmarried girl with complaints of cyclical lower abdominal pain and constipation was referred to Yüzüncü Yıl University Hospital in February 2005. Pelvic examination of the patient revealed an abdominal mass and imperforate hymen with normal secondary sexual characteristics. In ultrasonographic examination, hematocolpos and hematometra with extremely thin walled uterus and bilateral normal ovaries were recorded. Given her family history, she had 6 sisters and 2 brothers. Her parents were consanguineous. She and her family live in a rural region and had no history of exposure to any environmental or teratogenic hazards. Her eldest sister was diagnosed to have imperforate hymen 7 years ago and a cruciate hymenectomy was undertaken. This sister was married one year after the procedure and had given birth to 3 children. Other 2 sisters at age of 12 and 10 years were amenorrheic. They were called for screening to exclude imperforate hymen. They had normal hymeneal anatomy and they had normal genital organs in ultrasonographic examination. Pedigree of the patient's family is shown in Figure 1.

A stellate incision was performed to drain chocolate colored fluid of hematocolpos (Figure 2). No attempts were made to evacuate the uterus not to endanger the extremely thin uterine wall. MRI examination revealed normal genitourinary system. The patient was discharged without any complications.

Discussion

Imperforate hymen, reported to be a sporadic and isolated anomaly of genital tract, may present with cyclical, intermittent abdominal pain, abdominal mass, urinary retention, and

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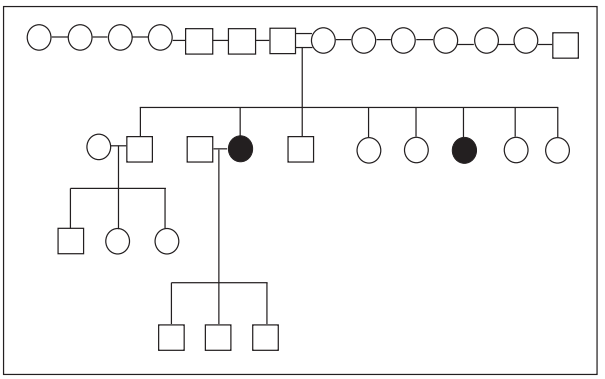

Figure 1. Pedigree of affected females in a consanguineous marriage.

constipation in pubertal girls. Hematometra and hematosalpinx may cause infertility, endometriosis and ectopic pregnancy in the future, therefore early diagnosis is important to avoid these complications.

Isolated cases of imperforate hymen can be due to phenocopy. These isolated cases could be a result of genetic cause (new dominant mutation, chromosomal anomaly, recessive condition, X-linked condition in males). Four cases of familial imperforate hymen are reported. McIlroy reported three sisters with imperforate hymen in one family, Usta et al. reported two families with three siblings from each family affected (1) and Lim et al. reported two sisters in one family. These cases were probably inherited by recessive mode of transmission (2). Generally diseases of autosomal recessive inheritance are suggested by individuals affected in a single sibling in one generation. On the other hand, a case of affected monozygotic twins and an affected daughter was suggesting dominant transmission (3). In our case, there were two affected sisters in a consanguineous marriage that suggests recessive

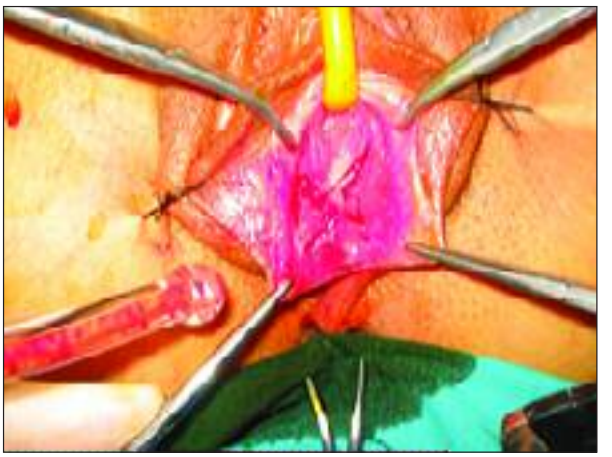

Figure 2. Stellate incision is made to drain hematocolpos and hematometra.

mode of transmission. Although presence of consanguity is not an absolute basis for autosomal recessive inheritance, it makes it more likely. Previous reports of familial imperforate hymen and our case suggest inheritance of imperforate hymen may be variable. Reports of familial imperforate hymen are limited to make a conclusion. However, it is of utmost importance to consider that imperforate hymen could be a familial condition and examination of family members is important to avoid late diagnosis and associated complications.

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