Bardet-Biedl syndrome associated with vaginal atresia: a case report

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This is a case report of Bardet-Biedl syndrome associated with vaginal atresia diagnosed in a 15-year-old girl. She had mild mental retardation; obesity; nistagmus, retinitis pigmentosa and optic atrophy in both eyes; accessory digit on the left hand; polydactyly in lower extremities; a mobile, painful, nonfixed mass of 6 cm in diameter in the pelvic region; a palpable cystic mass in front of the rectal wall; and no vaginal opening. Secondary sex characteristics were determined. The vaginal atresia was distinguished from vaginal agenesis by the presence of proximal vagina in radiological examination.

Key words: Bardet-Biedl syndrome, vaginal atresia.

Bardet-Biedl syndrome (BBS) was formerly grouped with Laurence-Moon-Biedl syndrome, but today is considered as a separate entity. It has autosomal recessive inheritance characterized by retinal dystrophy or pigmentary retinopathy, syndactyly, brachydactyly or polydactyly, obesity, mental retardation, hypogenitalism in males, amenorrhea in females, and structural abnormalities or functional impairment of kidney1-4.

Females with BBS have structural genital abnormalities, including persistent urogenital sinus, ectopic urethra, hypoplasia of the uterus, ovaries and fallopian tubes, uterus duplex, and septate vagina. Only a few patients with vaginal atresia and BBS have been reported to date2-5.7. In this study, we present and discuss a case of BBS associated with vaginal atresia.

Case Report

A 15-year-old girl presented with abdominal pain and distention continuing for a month. Her history revealed that her parents were close relatives and that she had a brother with mental retardation, blindness and hypogonadotropic hypogonadism.

At admission, her weight and height were 95% and 40% for age, respectively. She had mild mental retardation and obesity. The ocular examination showed nistagmus, retinitis pigmentosa and optic atrophy in both eyes. She had accessory digit on the left hand and polydactyly in lower extremities. Secondary sex characteristics consisting of breast enlargement, pubic hair, and enlargement of labia and clitoris were detected. Abdominal examination showed mild distention and tenderness. A mass of 6 cm in diameter, which was mobile, painful, nonfixed and with a smooth surface was found in the pelvic region. There was no vaginal opening. Rectal examination indicated palpable cystic mass in front of the rectal wall. Laboratory investigation results, including whole blood count, and liver and renal function tests, were within normal limits. Urinalysis and urine cultures were normal. The hormone levels were as follows: luteinizing hormone (LH) 3.21 mlU/ml; follicle-stimulating hormone (FSH) 6.58 mlU/ml; estradiol (E2) 18 ng/ml; progesterone 0.39 ng/ml; and testosterone 23.6 ng/dl (normal ranges according to age: LH 0.4-11.7<mlU/ml; FSH 1-9.2 mlU/ml; E2 3.4-17 ng/dl; progesterone 0.33-2 ng/ml; testosterone 20-38 ng/dl). Abdominopelvic ultrasonography showed an infraumbilical mass of 6 cm in size with fluid in the pouch of Douglas. Contrast-enhanced axial computerized tomography (CT) scan showed a urinary bladder fluid-air level and fluid-filled uterus, together with...
ovaries having well-marginated cystic areas (black arrows) (Fig. 1). The thickness of the myometrium was evaluated as normal in the fundus, but thinner in the corpus. The proximal vaginal blind pouch (black arrow) including dense fluid extended to the inferior of the symphysis pubis and was located on anterior side of rectum (Fig. 2). Bilaterally, kidneys were normal. Pelvic ultrasonography and CT findings revealed distal vaginal atresia.

The patient underwent surgery through a perineal approach. After proximal mobilization, vaginal pull-through was performed. Postoperative period was uneventful. She had normal menstruation periods, postoperatively.

Discussion

The BBS is an autosomal recessive disorder with locus heterogeneity. None of the responsible genes have been identified. The brother born to the consanguineous parents is homozygous.

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Fig. 1. The appearance of the urinary bladder, fluid-filled uterus and ovaries, including well-marginated cystic areas (black arrows) on computerized tomography.

Fig. 2. The appearance of the vaginal pouch (black arrow) extending to the inferior of the symphysis pubis on computerized tomography.
The Laurence-Moon-Biedl syndrome has been reported in identical twins. Our patient and her brother both had BBS.

Vaginal atresia may occur as a component of a few syndromes such as Mayer-Rokitansky-Küster-Hawser, Robinow and McKusick-Kaufman syndromes. Robinow syndrome has cardinal features including short stature, mesomelic shortening of forearms, frontal bossing, hypertelorism, antverted nares, triangular mouth, hypoplastic genitalia and vertebral and costal anomalies. McKusick-Kaufman syndrome (MKKS) is characterized by vaginal atresia with hydrometrocolpos, postaxial polydactyly, and congenital heart disease, and is also inherited in an autosomal recessive manner. It is more frequent in females. Our case had pigmentary retinopathy and obesity, which are additional findings when compared with MKKS.

The most common feature of BBS is retinal dystrophy. Nystagmus may be seen. Prognosis of visual function is poor and the fundal features are atypical and varying. A study including 109 BBS patients and their families has shown that the average age of diagnosis is nine years, postaxial polydactyly is present in 69% of patients, obesity begins to develop at around 2-3 years, and retinal degeneration is not apparent until a mean age of 8.5 years. Hypogonadism is a cardinal feature in Laurence-Moon-Bardet Biedl (LMMB) and BBS and is more common in males. Renal abnormalities are characteristic but not in all cases as in our patient. Facial dysmorphism, behavioral traits, neurological, speech and language deficits, and dental anomalies had all been reported recently. These features may facilitate earlier diagnosis of this disorder.

The diagnosis of BBS is rarely established in early childhood. The association of atresia of the vagina and other malformations of female genital structures in individuals with BBS is often not manifested in childhood and should be looked for carefully. Prognosis of distal vaginal atresia is excellent. The diagnosis of high or low-type vaginal atresia is mandatory. Although modern imaging methods may be helpful, the most important step is a physical examination. The aim of treatment is distal vaginal drainage, which can be achieved by perineal procedure in most cases. Laparotomy is indicated only in cases of high vaginal atresia.

In conclusion, vaginal atresia must be remembered as one of the cardinal findings in BBS.

REFERENCES

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