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Case Report

Laparoscopic Management of Ovarian Cysts Torsion in the Mccune-Albright Syndrome

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Abstract:

A 10-year-old girl with McCune-Albright syndrome, a rare genetic disease, presented with ovarian cysts torsion when entering the emergency room. She had a past medical history significant for precocious puberty, café-au-lait spots, and fibrous dysplasia associated with McCune-Albright syndrome.

Previous episodes of recurrent vaginal bleeding, precocious puberty, and ovarian cysts were clinically managed with an alternating therapy of tamoxifen or aromatase inhibitors to alleviate symptoms. Preoperative tumor marker tests were negative, showing no evidence of malignancy; therefore, laparoscopic surgery was adopted.

During the procedure, the twisted ovary was repositioned and the lesion resected. The ovary was ultimately preserved, different from previous reports of inadvertent oophorectomy in prepubertal girls with MAS.

Postoperative pathology confirmed a benign ovarian cyst. The child recovered well, with no recurrence observed during a 2-year follow-up. Menstruation returned regularly under pharmacological control.

Key words: McCune-albright syndrome; fibrous dysplasia; café-au-lait spots; precocious puberty; ovarian cysts torsion

Case Presentation

A 10 - year - old girl with McCune - Albright syndrome was hospitalized in our department due to abdominal pain symptoms. The patient had a history of vaginal bleeding, which started six months after her first birthday. She was also presented with a distinctive combination of fibrous dysplasia (FD) of bone, café - au - lait (CAL) skin spots (the Café - au - lait spots could be seen on the skin of the chest, back, and thighs during physical examination), and hyperfunctioning endocrinopathies with ovarian cysts infrequently recurrence. Genetic testing for GNAS mutations had been carried out when 4 years old. Letrozole and tamoxifen alternately was used for the management of endocrine disorders with recurrent ovarian cysts.

A pelvic ultrasound was performed when the girl was admitted to the emergency room, which showed an ovarian cyst of the right ovary measuring 6.9 cm × 5.9 cm × 4.0 cm with a mixed - echo mass inside, and there was a visible echo mass of 3.7×3.5×3.3 cm at the same time of administration (Figure.1-2).

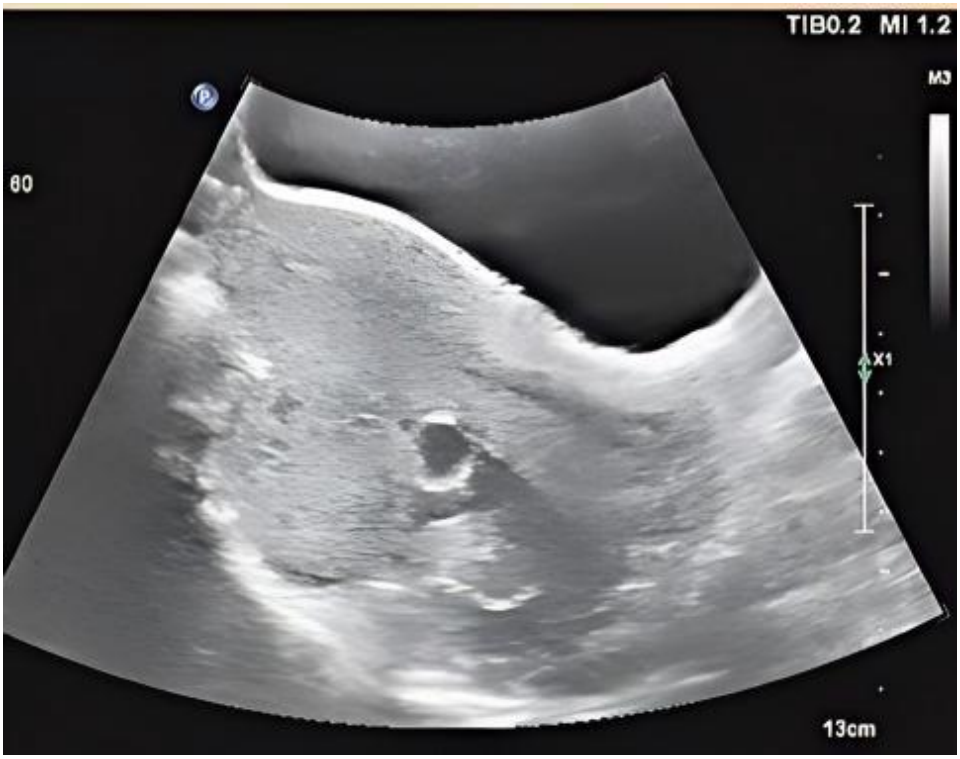

Figure 1: Pelvic ultrasound image. a ovary cyst, measuring 6 cm in diameter.

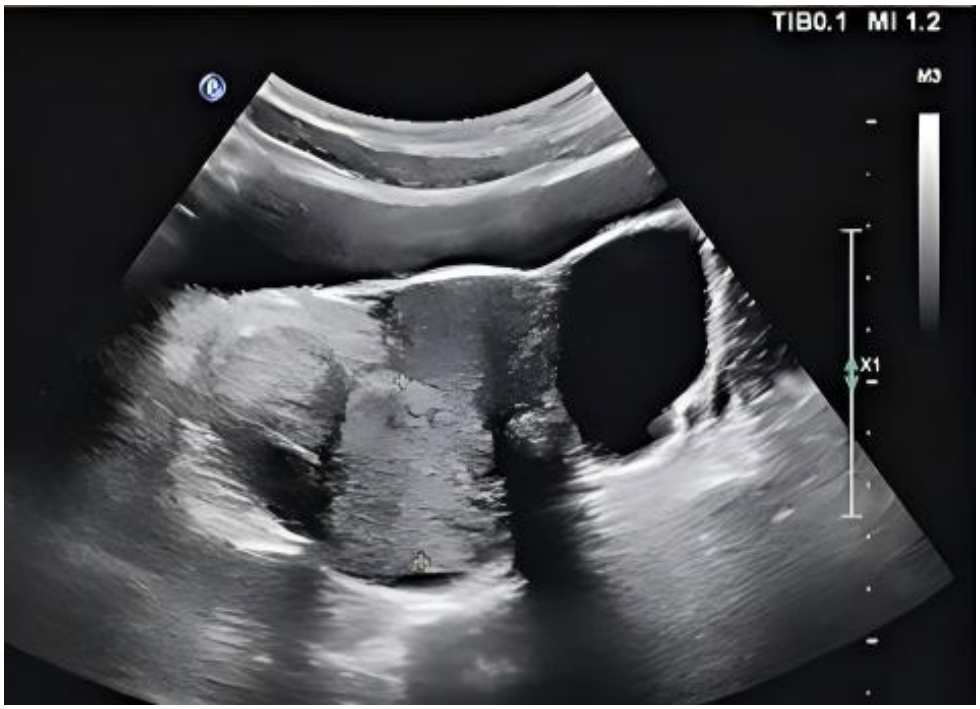

Figure 2: Representative pelvic ultrasound images. Dynamic monitoring of ovarian cyst torsion.

Adjacent to it, another heterogeneous high - echo of 4×3.9×2.9 cm could be seen, which ran in a "spiral" pattern and changed in the cross - section in a "vortex" pattern. CDFI showed that no obvious blood - flow signal was observed inside the clear content and a thin layer. A left - sided cystic ovarian lesion was demonstrated, which measured 5 cm x 5.1 cm x 4.0 cm without internal color Doppler flow. The physician suspects that the ovarian cyst has torsaded according to the ultrasound findings.

All examinations were completed sequentially during the hospitalization. Laboratory investigations revealed elevated levels of estradiol (242 pg/mL), FSH at 1.11 mUI/mL, and LH levels at 0.7 mUI/mL, along with normal testosterone (1.42 nmol/L) and prolactin (11.43 ng/mL). No abnormalities were detected in tumor markers such as CA125. Magnetic resonance imaging of the brain and pituitary showed no remarkable findings. Otherwise, a torsion lesion on the ovary was revealed (Figure. 3, 4).

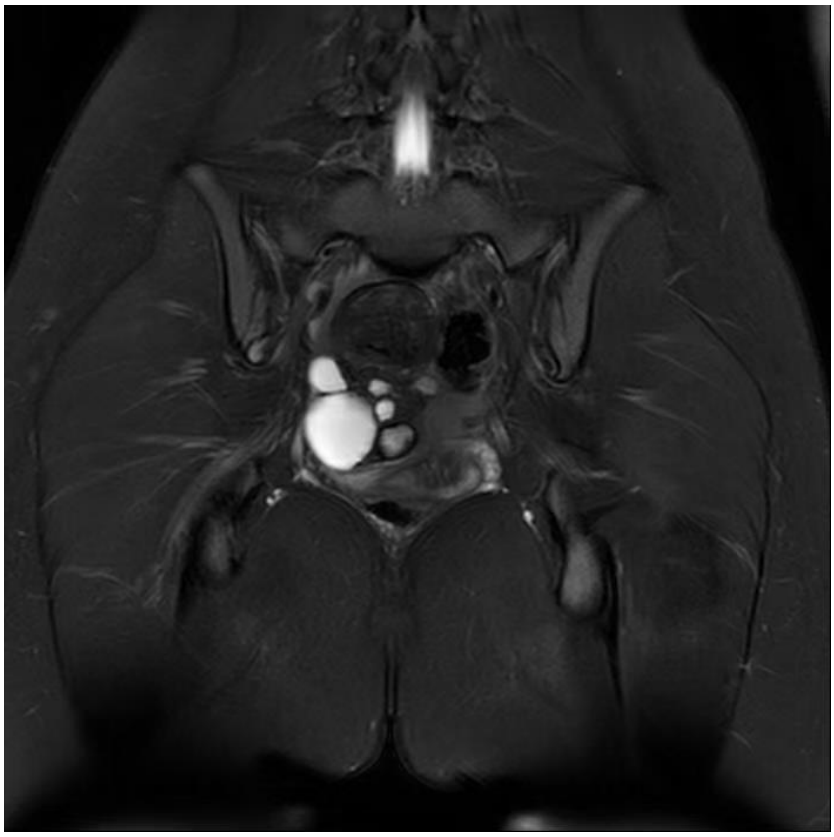

Figure 3: MRI scan of the pelvis showing the right ovary cystic ovary torsion lesion.

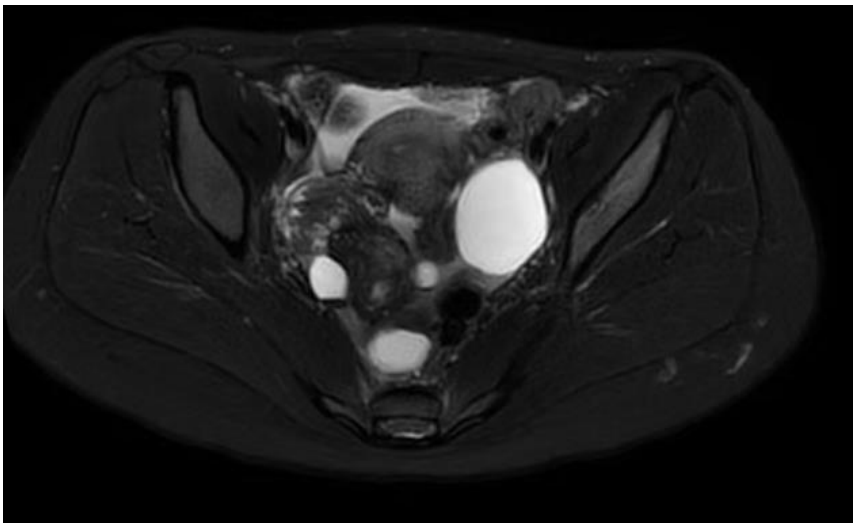

Figure 4: MRI coronal scan of the pelvis showing the right ovary cystic ovary torsion.

On skeletal imaging, there were multiple lytic and sclerotic lesions of the distal radius, as well as fibrous dysplasia (FD) with a "copper - beaten" skull.

The departments of Pediatrics, Endocrinology, Obstetrics and Gynecology, Pathology, Imaging, and Anesthesiology and Surgery jointly conducted an assessment and unanimously agreed that laparoscopic surgery was feasible. An emergency laparoscopic surgery was performed after the discussion. During the procedure, a torsion of the right ovarian cyst pedicle with necrotic changes was observed (Figure. 5).

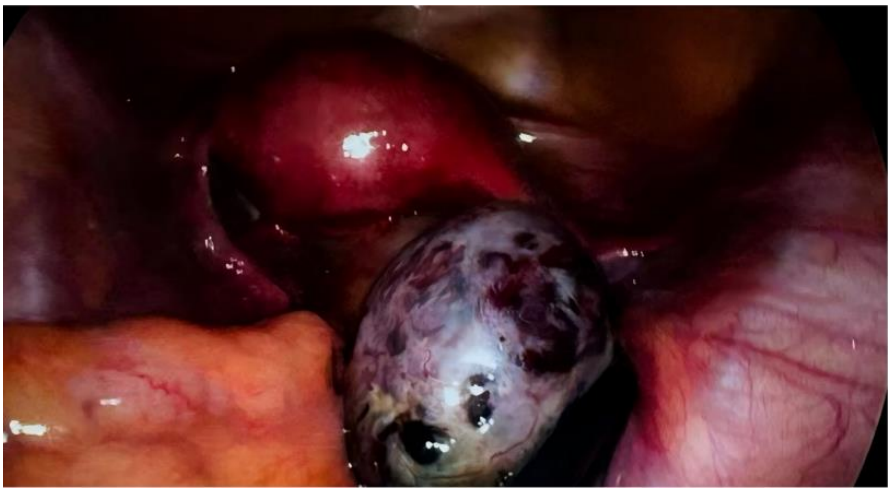

Figure 5: Ovarian cyst torsion on laparoscopy

A 4 - cm ovarian cyst on the left side, with a translucent appearance, was subsequently excised. The position was restored, followed by warm - saline immersion to restore blood supply, and finally, part of the ovary was successfully preserved. The cyst was excised while maintaining ovarian function. The girl had a favorable recovery postoperatively.

Surgical pathology revealed benign ovarian cysts with hemorrhagic necrosis. The child recovered well, with no recurrence observed during a 2 - year follow - up. Menstruation returned regularly under pharmacological control.

Discussion

McCune - Albright syndrome (MAS) is a rare disorder, occurring in 1 in 100,000 to 1 in 1,000,000 live births [1]. It is marked by the most common endocrine manifestations of fibrous dysplasia (FD) of bone, café - au - lait (CAL) skin spots, and hyperfunctioning endocrinopathies. It is more common in girls than in boys [2]. Typically, vaginal bleeding or spotting is followed by the development of breast tissue.

In our case, the patient was reported to have significant vaginal bleeding, café - au - lait (CAL) skin spots, precocious puberty but without bone pain at the time of presentation. Precocious puberty in females is caused by elevated serum estradiol levels due to intermittent autonomous activation of the ovaries. These symptoms occur because of the activation of gonadotropin receptors in the ovaries, despite the absence of gonadotropin stimulation. This leads to ovarian enlargement, cyst formation, and the secretion of estradiol, which triggers early puberty and abnormal vaginal bleeding. The girl had vaginal bleeding at age 6 months and recurrent ovarian cysts in this case.

Diagnosis is primarily clinical, requiring at least 2 features of the triad, supported by hormonal tests and imaging. Genetic testing for GNAS mutations can confirm the diagnosis but is not always necessary [3,4]. The patient was diagnosed with a gene mutation in the medical history and began endocrine therapy. Letrozole and or Tamoxifen was used for the management in endocrine disorders.

The Guanine Nucleotide - binding protein, Alpha - stimulating activity polypeptide (GNAS) gene encodes for the ubiquitously expressed stimulatory subunit alpha of the G protein (Gsα). G protein couples hormone receptors to adenyl cyclase, which is necessary for the generation of intracellular cAMP that mediates G - protein - coupled hormone signaling. The reported GNAS mutations in Arg 201 and Gln 227 in McCune - Albright syndrome inhibit the guanosine triphosphate hydrolase (GTPase) catalytic ability of Gsα, making it impossible to control the Gsα activation[3]. As a result, there is excessive cAMP production, even in the absence of stimulating hormones. Caused by post - zygotic somatic mutations in the GNAS gene, this cascade of early estrogen exposure may lead to precocious puberty and ovarian cysts.

The treatment of MAS is rather complex and challenging. Clinically, aromatase inhibitors and estrogen receptor antagonists are usually used to reduce vaginal bleeding and alleviate symptoms of precocious puberty such as breast development [5,6]. Bisphosphonates and other drugs are used to relieve bone pain. However, all the above - mentioned drugs are off – label [6]. Before treatment, it is necessary to communicate fully with the parent(s) of the child and/or the patient. It is also recommended to refer the patient to a pediatric endocrinology specialist with treatment experience.

The management of multi - system lesions (such as pituitary tumors, hepatobiliary system tumors, etc.) or ovarian cyst torsion requires the participation of multiple disciplines and long - term follow - up and management. Undoubtedly, the presence of an ovarian cyst requires a multidisciplinary approach with close collaboration among pediatricians, endocrinologists, surgeons, and pediatric gynecologists to prevent unnecessary cystectomy or oophorectomy [6]. In the past, a small number of cases of MAS patients who underwent “mistaken identity” oophorectomy have been published [7-9], thereby emphasizing the importance of timely MAS diagnosis through fruitful collaboration among different specialties.

Laparoscopic surgery is the main therapeutic strategy for large and persistent simple ovarian cysts in childhood, particularly with torsion of ovarian cysts[10-12]. Surgery is currently adopted as the gold standard for the diagnosis of ovarian mass torsion, which can achieve to determine the degree of torsion and determine tissue activity, in which the timing of surgery is rather critical. Finally, the operation was performed under the general anesthesia. During the operation, we found a cyst about 7 cmx 7 cmx6 cm, rotate 360 degrees counterclockwise. The torsion ovary was reset. The lesion was finally removed from the side incision and the incision was sutured. Some findings suggest that unilateral oophorectomy may be helpful for the treatment of MAS - associated infertility in select cases [13-16]. However, this approach should be undertaken with extreme caution, considering the potential for harm to fertility, and only under the supervision of an experienced reproductive specialist. Some report and review revealed that the reproductive function and fertility in girls with MAS are normal. The significant difference comparison with previously reported cases, we determined to preserve ovarian function and fertility preservation, in our case, after multidisciplinary discussion. Increased awareness of this complex disease among all pediatric practitioners is

needed to prevent inadvertent oophorectomy in prepubertal girls with MAS.

Conclusion

In summary, we described a 10 - year - old girl with an estrogen - producing ovarian cyst unfortunately torsion, which, along with a café au lait skin macule, is all suggestive of MAS diagnosis. We highlight the importance of careful assessment and strive to preserve the reproductive function of adolescents.

Conflicts of Interest

The authors declare no conflict of interest.

Funding Declaration

There was no Funding.

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