A rare presentation of Peutz-Jeghers syndrome in children

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Introduction

The sex cord tumor with annular tubules (SCTAT) is a rare tumor associated with Peutz-Jeghers Syndrome (PJS) in 36% of cases [1]. Most PJS associated SCTAT tumors are benign but 20% of sporadic cases are malignant [1]. The mean age of presentation PJS is 27 years [1]. It is rare in children and the presentation is with gonadotrophin independent precocious puberty (GIPP) [1]. Due to its rarity, there is no standardized management established [2]. Recurrence in SCTAT in children with PJS is not reported [1]. However, sporadic SCTAT has the potential to recur [1,2].

Case report

A four and half-year-old girl was referred due to 3 episodes of cyclical vaginal bleeding over a period of four months. There was gradual breast enlargement and rapid increment in height over seven months.

She was the only child in the family of non-consanguineous parents, who was born at term. There was no history of precocious puberty in the family but the maternal uncle was diagnosed, patient with the PJS and was regularly followed up at the surgical unit.

She was an averagely built girl with height and weight falling in between – 1SD to median centiles. Her vital parameters were unremarkable. There were melanocytic macules on the lips without syndromic features. Her breast stage was Tanner II-III, pubic and axillary hair stage was Tanner I-II. Her bone age was advanced at 6 to 7 years and Leutinizing Hormone was 0.09 IU/l (pre-pubertal <0.2 IU/L) with Follicular Stimulating Hormone of 0.05 IU/l (pre-pubertal 0-4 IU/L). The estradiol level was 581pmol/l (2.7- 27.8 pmol/l). Her beta hCG and alpha feto proteins were normal.

The MRI brain was normal and the pituitary was compatible with the pre-pubertal stage. The ultrasound scan of the pelvis revealed a pubertal uterus with an endometrial thickness of 10mm without ovarian lesions. We started her on letrozole as it is an aromatase inhibitor, so it blocks the conversion of androgens produces by the ovaries into estrogen, thereby preventing the progression of puberty. We followed up the child with ultrasound scans every 6 months. After one and half years, her ultrasound scan revealed a mass lesion in the left ovary. An MRI was performed which showed well defined left ovarian solid lesion with post-contrast enhancement. She underwent a left side salphingo-oophorectomy at which a tumor measuring 20×20×15mm was excised. It was well localized with no metastasis. The histology report revealed an ovarian SCTAT. Considering the PJS, we proceeded with upper and lower endoscopy which showed polyps in the antrum of the stomach and the duodenum. She was planned to undergo a repeat endoscopy in 10 years and regular follow-up was arranged.

Discussion

The SCTAT is a distinctive ovarian neoplasm first described by Scully [3]. Its morphology is an intermediate between the granulosa cell tumor and the Sertoli cell tumor with the potential to differentiate focally either into granulosa or Sertoli cell tumor[3]. There is a strong association with SCTAT and PJS, wherein in a study, one-third of patients were diagnosed with the lesion [3]. It is broadly classified as SCTAT with PJS and without PJS [3]. The SCAT associated with PJS is a hematoma; benign, multifocal, bilateral, very small size or microscopic and calcified lesion [3]. The clinical presentation is due to estrogen and progesterone secretion like menorrhagia, post-menopausal bleeding, precocious puberty, and sterility [3]. They underwent surgery and recurrence was not reported. A single case report of malignant SCTAT with PJS and recurrence was reported in a 47-year-old lady [4].

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PJS has an incidence of 1 in 25,000-300,000 and is a result of a mutation in the STK 11/LKB 1 gene which is inherited in an autosomal dominant manner [4]. Approximately 60-78% have a relative affected by the condition. The main clinical criteria for the diagnosis are family history, mucocutaneous lesions, and hamartomatous polyps mainly in the gastrointestinal tract. The melanocytic lesion appears before the age of 5 years and common sites are lips, oral mucosa, and the skin [5]. In the pediatric age group, common presentation is with intussusception and bowel obstruction around the age of 8 years [5]. Our patient had characteristic mucocutaneous pigmentation, polyps in the small bowel, and the stomach with a family history of Peutz Jegher syndrome. However, our patient did not have a history of bowel symptoms.

Conclusion

Children presenting with GIPP need proper assessment and meticulous follow-up as it could be a manifestation of a gonadal tumor. Peutz-Jeghers is one of the causes for GIPP and patients should be investigated if history and clinical features are suggestive of the condition.

Authors' contributions

DSG, BCL, CN and NA were involved in the clinical management of the patient and DSG did the drafting of the manuscript. NA critically evaluated and revised the draft. SG evaluated the biopsy specimen and submitted the report. All authors approved the final submission

Disclosure statement

We have no conflict of interest to declare.

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Patient consent

All the information regarding the patient was completely anonymized. We have obtained the written consent for medical photography from the mother of this child.

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