Unusual ovary formation in a girl with McCune-Albright syndrome

Makazan N.[1], Orlova E.[1], ., Artemova A.[1], Vladimirova V[1], Vorontsov A.[1], Kareva M.[1], Peterkova V.[1]

[1] Endocrinology Research Centre, Moscow, Russia

Introduction

McCune-Albright syndrome (MAS) is a rare disorder caused by somatic mutations in *GNAS* gene leading to fibrous dysplasia (FD), cafe-au-lait spots and hyperfunctioning endocrinopathies. The common feature of MAS in girls is peripheral precocious puberty (PP) with recurrent ovary cysts. Few cases of ovary tumors have been described to date.

Case Description

8.5 year-old girl with MAS:

Diagnosis established at the age of 4.8 (FD of the left lower limb, cafe-at-late spots)

Annual screening for components of MAS: blood tests to reveal additional endocrine features; head CT, X-ray, ultrasound of the thyroid and gonads.

From 4.8 to 8.5 years old: 3 episodes of vaginal bleeding with recurrent ovary cysts (persisting for the period no longer than a month). No additional components were revealed so far.

During the evaluation at the age of 8.5:

- 1st ultrasound: signs of solid l lesion with cystic zones of the right ovary.
- 1st MRI: large ovarian lesion (4.5x4.3cm) with small amount of fluid (ascites) in the pelvis - **disgerminoma (?)** (fig.1).
- Blood test at that time: estradiol 63 pmol/l, LH 0,216 U/l, FSH 1,06 U/l, HE4 40 pmol/l (<70), CA125 12.5 U/ml (0 – 30,2)
- 2nd MRI in 9 days: **significant decrease in the lesion size** and disappearing of the ascites (fig.2)
- At 4 months follow up the lesion in the right ovary has been **completely regressed** (fig.3)

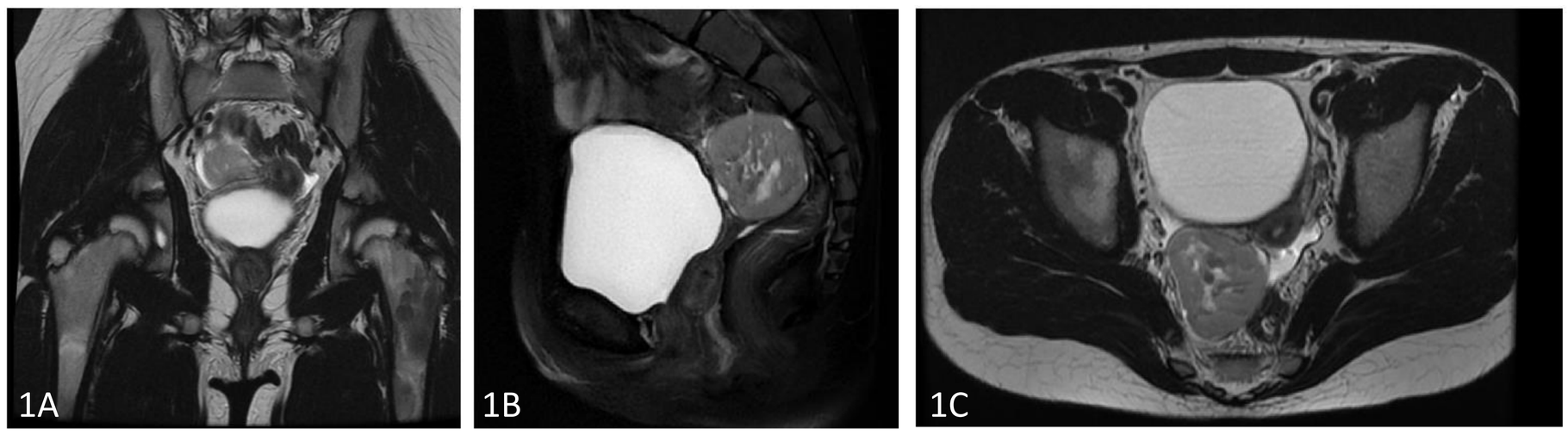

Fig.1A-1C MRI of the pelvis 25.03.19: MRI-signs suspective for disgerminoma

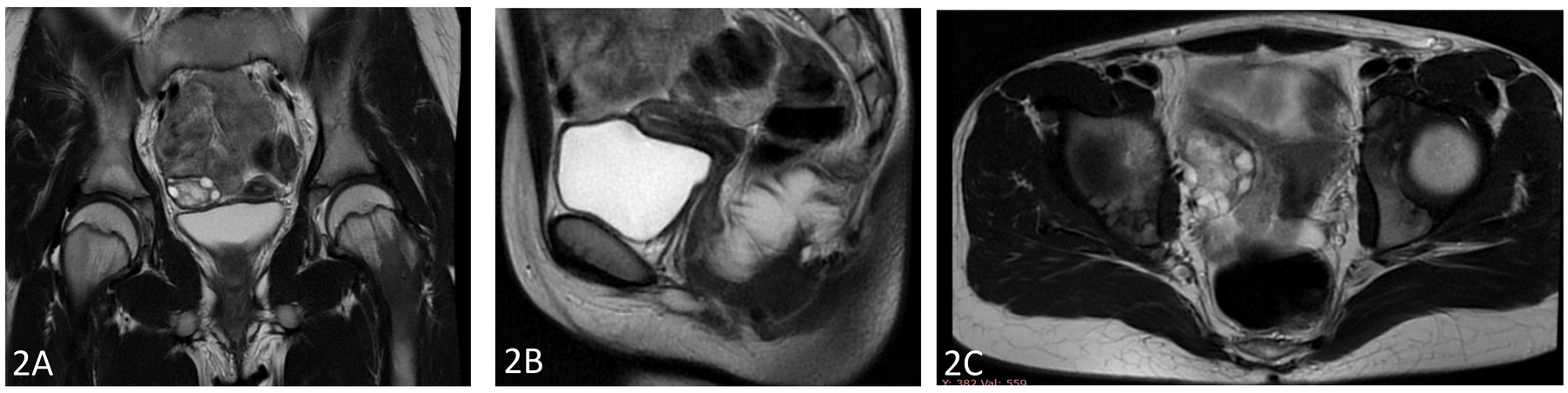

Fig. 2A-AC MRI of the pelvis 04.04.2019 significant decrease in size of the lesion and disappearing of the ascites

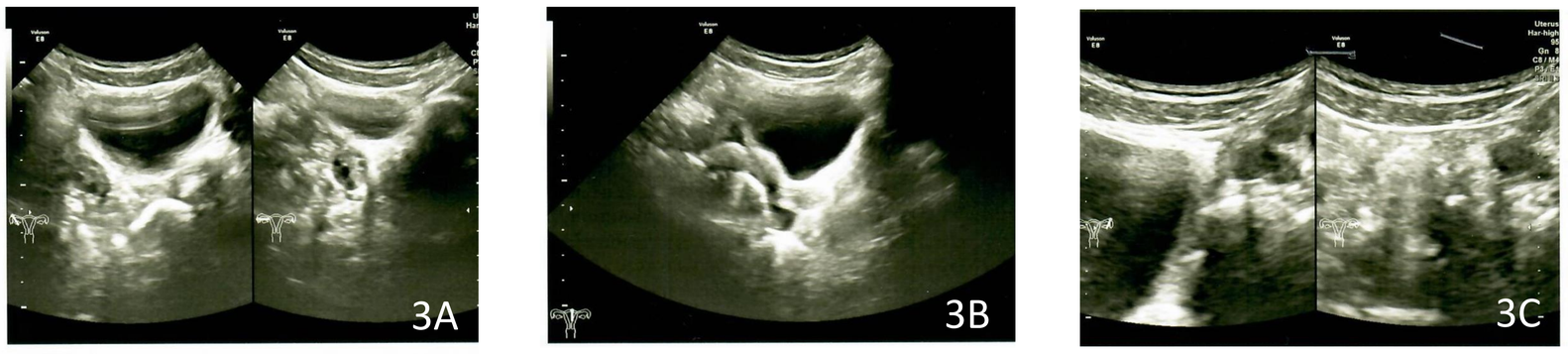

Fig.3A-3C Ultrasound of the pelvis: no signs of the ovarian lesion.

Conclusion

In MAS patients it can be difficult to establish treatment approach to the ovary lessons, considering tendency to ovary cysts on the one hand and possibility of developing ovary tumors on the other.