CONGENITAL ADRENAL HYPERPLASIA AND MULTIPLE SCLEROSIS – COINCIDENCE OR NOT?

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Introduction

Congenital adrenal hyperplasia (CAH) is an inherited recessive disorder of adrenal steroidogenesis, generally caused by total or partial deficiency in 21-hydroxylase, due to deletions or mutations of CYP21 gene.

Some studies suggest that the association between CAH and Multiple Sclerosis (MS) could be nonincidental: a possible MS susceptibility locus is on chromosome 6p21, on which the CYP21 gene is located.

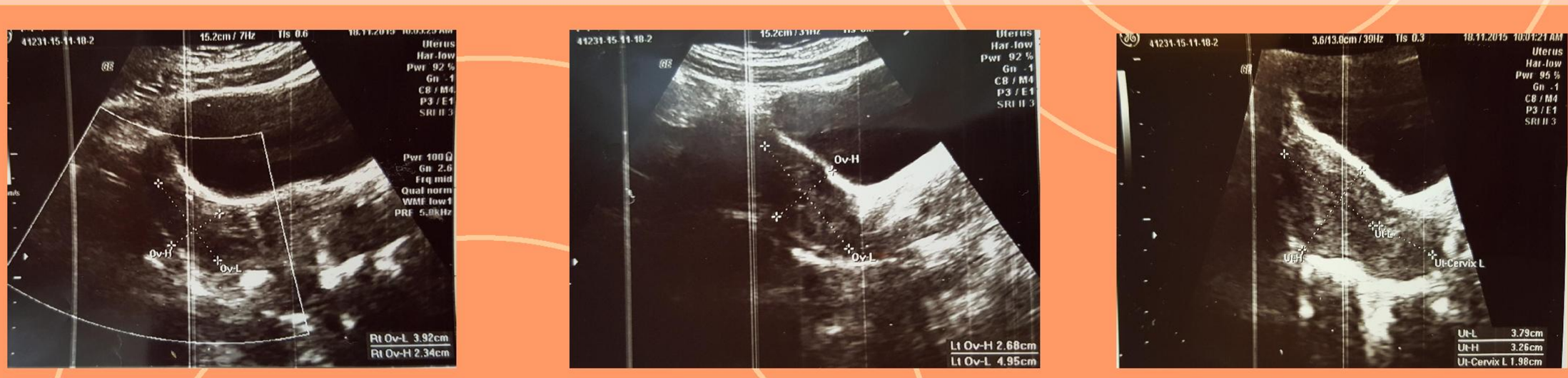
Case report

We report the case of a 16-year-old female who presented to our clinic in November 2015 with complaints of severe hirsutism exacerbated during puberty, diplopia, vertigo. In April 2014 the patient was treated for hirsutism with Spironolactone 75mg/day but she developed nausea, vomit, ataxia and the treatment was stopped after one month. The patient recovered spontaneously after a few weeks but diplopia and vertigo appeared one year later.

Physical examination revealed overweight (BMI=29,4kg/m2), severe hirsutism (Ferriman Gallwey score=24), normal pubertal development, regular menses, no genital anomalies. Laboratory tests: Estradiole=43.66pg/ml, FSH=5,84mIU/ml, LH=3.08mIU/ml, Cortisol-8am=27.8mcg/dl, ACTH=56.5pg/ml, Cortisol-8am after 1mgDXMovernight=1.41mcg/dl, DHEAS=438.6mcg/dl(65-368), Testosterone=46.72ng/dl, 17OH-progesterone=2.28ng/ml, 17OH-progesterone after ACTH stimulation test=21ng/ml, normal TSH, ATPO, FT4, Prolactine. Abdominal ultrasound was normal. Ophthalmologic evaluation: horizontal nystagmus, papilledema, sixth nerve paresis. Brain IRM revealed several T2-hyperintense white matter lesions, located supra-and infratentorial maximum size 2.4cm.

Non-classic CAH and MS were diagnosed and the patient was referred to the pediatric neurology department.

The association between oral contraceptive (OC) and Spironolactone is probably the best treatment for hirsutism although there are contradictory data regarding the use of OC in MS patients.



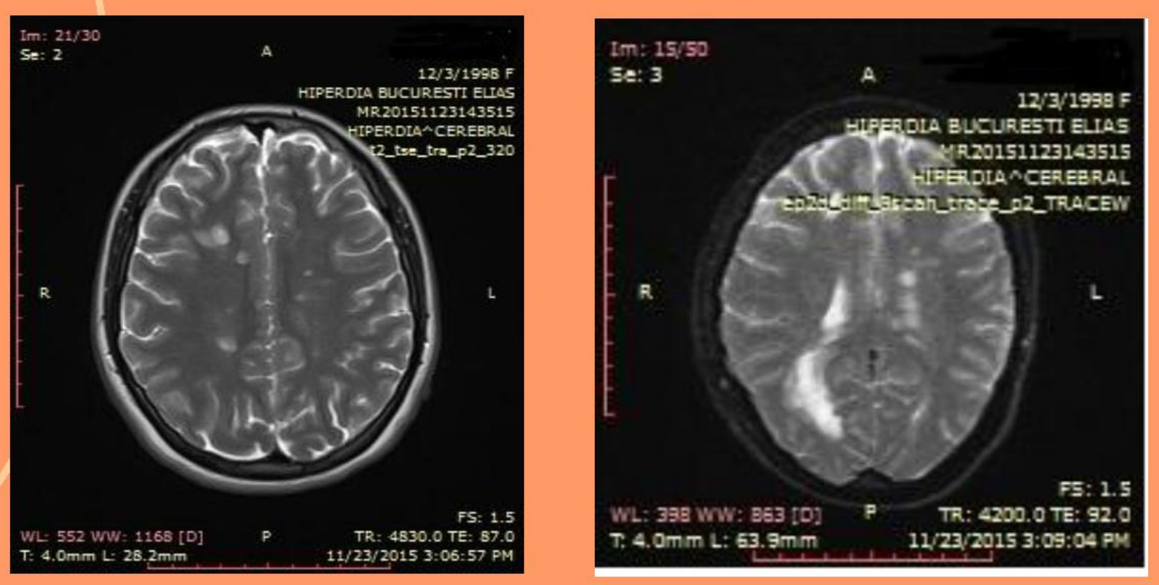
Trans-abdominal ultrasound of the uterus and ovaries

Conclusions

To the best of our knowledge this is the second case of CAH associated with MS to be reported in the literature.

Further extensive epidemiological and genetic studies could explain the relationships between MS and CAH.

Endocrinologists should search for neurological signs and symptoms in CAH patients and neurologists should recognize the presence of CAH in MS patients.



Brain IRM

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