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Letter to the Editor

Polycystic ovary syndrome: A new phenotype in mosaic variegated aneuploidy syndrome?

Syndrome des ovaires polykystiques : un nouveau phénotype du syndrome d'aneuploidie en mosaïque ?

1. Clinical report

A 27-year-old woman with a history of short stature in infancy and oligo menorrhea presented with secondary amenorrhea. She was the product of a third pregnancy of non-consanguineous parents. She had no cognitive impairment. She had her menarche at the age of 12 years, since then she reported chronic menstrual irregularities. She has had secondary amenorrhea since twelve months. Physical examination showed a short neck without microcephaly, short stature with a height of 1.22 meters and a short proximal limb segments (Fig. 1). Her weight was 45 kg and body mass index 30 kg/m^2 , with a waist circumference of 80 cm. Systolic blood pressure was 90 mmHg and diastolic blood pressure was 60 mmHg. She had a facial dysmorphia: temporal bossing, triangular face, hypertelorism and micrognathia. She had male pattern with a frontal baldness without hirsutism or other signs of virilism (Fig. 2). Skeletal abnormalities were knee varus, short fingers and toes, broad first toe and a wide sandal gap between the first and the second toes (Figs. 3 and 4).

The pituitary hormone tests revealed normal baseline gonadotropin level. The progesterone withdrawal test was



Fig. 2. Facial dysmorphia: short neck, temporal bossing, micrognathia.



Fig. 3. Feet skeletal abnormalities: broad first toe and a widely sandal gap.



Fig. 1. Patient's morphotype: short stature short proximal limb segments.



Fig. 4. Hand skeletal abnormalities: short fingers.

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Table 1
Hormonal tests.

	Reference range	2010	2011	2012
FSH (UI/L)	1–20	4.8	9.3	
LH (UI/L)	2–20	4.7	7.5	
Estradiol (pg/mL)	39–189		20	84.2
Prolactin (ng/mL)	2–25	11	9	
FT4 (ng/dL)	0.71–1.85	1.32	1.14	
TSH UI/mL	0.12–3.4	1.86	2.48	
Cortisol baseline (μg/L)	42–384	110	215	
Testosterone (ng/mL)	0.1–0.58	0.75	1.05	
IGF-1 (μg/L)	108–247		187	

Table 2
Metabolic tests.

	Reference range	June 2011	November 2011
Fasting glucose (g/L)	0.7–1	1.15	
Plasmatic glucose 2 hours after OGTT ^a			3.44
Total cholesterol (g/L)	1.2–2	2.16	2.28
Triglyceride (g/L)	0.5–2	1.20	1.41
HDL cholesterol (g/L)	0.35–7	0.35	–

^a OGTT: oral glucose tolerance test: the glucose cut-off for 2h-75 g OGTT was 2 g/L.

positive. The insulin tolerance test showed an insufficient GH response (5.3 ng/mL). IGF1 level was normal for age. Thyroid and adrenal hormones were normal. Testosterone level was moderately elevated (Table 1). The metabolic tests showed hypercholesterolemia, low HDL cholesterol and a type 2 diabetic status (Table 2). The pelvic ultrasonography revealed normal ovary volume: 6.7 mL on the right, 8.4 mL on the left and multiple follicles in both ovaries. Pituitary MRI revealed a hypoplastic anterior pituitary gland and a Rathke's cleft cyst (Figs. 5 and 6).



Fig. 5. Coronal MRI showing hypoplastic anterior pituitary.

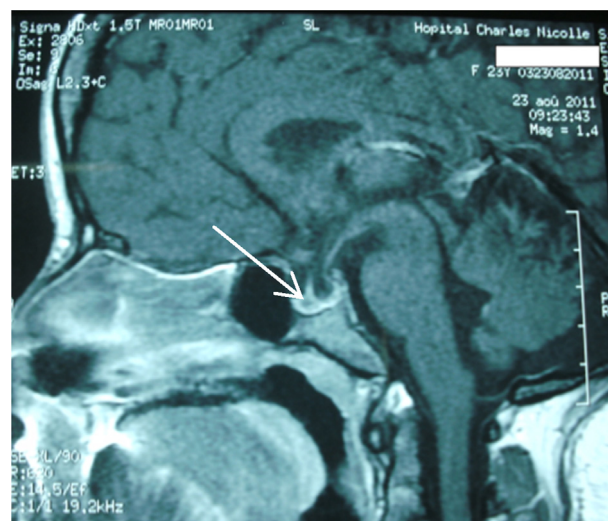


Fig. 6. Gadolinium-enhanced sagittal T1 MRI showing Rathke's cleft cyst (white arrow).

The initial peripheral blood chromosome study showed multiple and diverse aneuploidy in 20% of mitotic cells. The second showed aneuploidy in 30% of mitosis (Fig. 7).

The screening for neoplasia was negative; full blood count was normal. Radiographs showed a normal bone skeleton, they did not show evidence for ovarian tumor. Abdominal pelvic ultrasound did not show evidence for ovarian tumor.

During 2 years follow-up, amenorrhea persisted but bleeding occurred whenever progestative therapy was used.

2. Discussion

Our case is the first report of metabolic syndrome and secondary amenorrhea in mosaic variegated aneuploidy syndrome (MVA). It is a very rare condition characterized by constitutional mosaic aneuploidies, non-specific phenotype including microcephaly, mild malformations, growth and mental retardation, and an increased risk of malignancy [1,2]. Dysmorphic facial features in MVA syndrome are low-set ears, micrognathia, epicanthic folds, occipital prominence, broad nasal bridge, triangular face, frontal bossing and hypertelorism. Other neurological and eye abnormalities, skeletal hand and foot abnormalities and dermatological anomalies have also been described [3].

The diagnosis of MVA syndrome is based on the increased rate of variable aneuploidies in observed cells. The proportion of aneuploid cells in individuals with MVA is variable, but is usually more than 10% and substantially greater than in normal individuals [4]. The phenotype was mild in our patient, including craniofacial dysmorphic feature and growth retardation, genetic study showed predominantly nonosomies with multiple trisomy in blood lymphocyte. The oligomenorrhea lets us discuss different diagnosis. The direct relationship between hypoplastic pituitary and secondary hypogonadism was less plausible as the lack of gonadotropin has not been described in MVA syndrome before and the progesterone withdrawal test was positive

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