A case of hypopituitarism in a patient with Cantù syndrome

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INTRODUCTION

Cantù syndrome (OMIM #239850) is a rare disorder characterized by:
- ❖ congenital hypertrichosis,
- ❖ neonatal macrosomia,
- ❖ a distinct osteochondrodysplasia,
- ❖ cardiomegaly ([1]).

Other findings described are:
- ❖ vascular abnormalities,
- ❖ pulmonary hypertension,
- ❖ generalized oedema at birth,
- ❖ mild learning disability
- ❖ behavioral problems and others ([2]).

GENOTYPE

The syndrome is related to an heterozygous pathogenic variant in *ABCC9* or *KCNJ8* ([3], [4]), which can be inherited in an autosomal dominant manner or due to a *de novo* mutation

chr12 (p12.1) | 12q12 | 15 | 22

CASE REPORT

We report a particular case of a 15 years old girl, affected by Cantù syndrome clinically diagnosed (genetic tests are currently being conducted), with hypopituitarism.

G. was born at 31+4 week of gestational age with caesarean section. During the pregnancy polidramnios and gestational diabetes (treated with insulin therapy) were evidenced. At birth she presented respiratory distress and she needed endotracheal intubation. Bronchopulmonary dysplasia was than diagnosed in the first moths of life. She presented also a patent ductus arteriosus, that needed surgically correction. She had a normal motorius development but a slight language and cognitive retard.

Physical examination: G. presents macrocephaly and typical coarse facial features with broad nasal bridge, anteverted nares, enlarged nasal filter, macroglossia and enlarged pinna. She also has hypertrichosis, vitiligo area on the sternum, ligamentous laxity, pectus carinatum and hoarsely voice.

Other manifestations: Bone RX showed multiple abnormalities: endosteal thinning of the humerus and widening of its diaphysis (Fig. 1 e 2); smooth metaphyseal cortical thickening of proximal humerus, distal femur, and proximal tibia; omeral osteochondrosis. Over the time she developed a compensated left ventricular dilatation (Fig. 4 chambers echocardiography). CGH array didn't show any alteration.
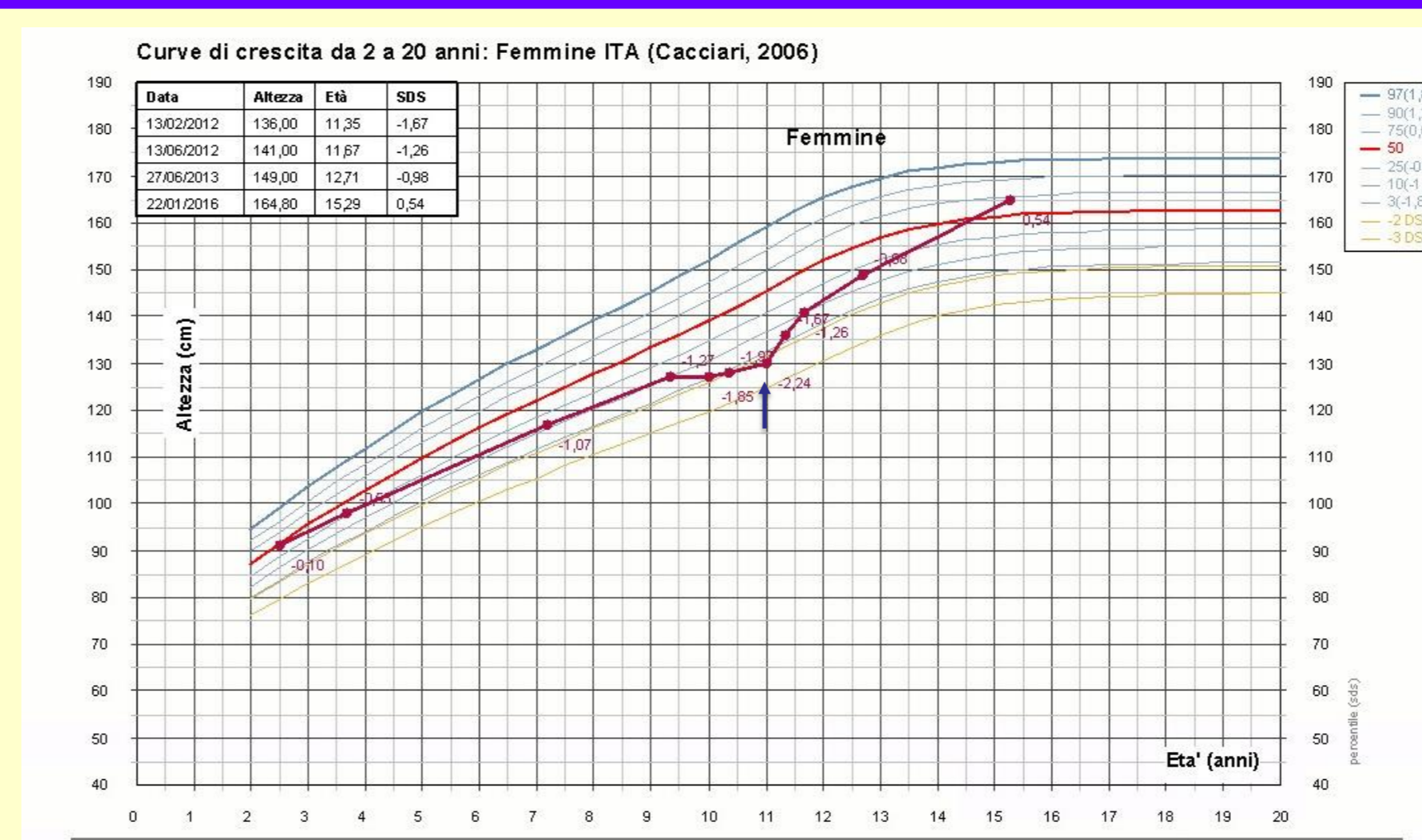

Fig.1 e 2

Fig. 3: Growth curve. The arrow indicates the beginning of GH therapy

Fig. 4

Endocrinological features: At birth a central hypothyroidism has been diagnosed and substitutive therapy was started; it was than stopped at 28 month of life in another hospital. At 11 years old, GHRH test detected a central GH deficiency and a recombinant therapy was set up (fig. 3). Subsequently the central hypotirodism became manifest again, thus needing L-tiroxine. Finally, for a delay in puberal development, at 14 years old we performed a GnRH test, that showed low FSH and LH levels, confirming an hypogonadotrophic hypogonadism, for which she's on estroprogestinic therapy. Furthermore MRI showed a pituitary hypoplasia.

CONCLUSIONS

We described a case of Cantù Syndrome with hypopituitarism, thus underlying the importance to assess, at the beginning, the endocrine status of those patients and to maintain a strict surveillance of hormonal axes during whole life..

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