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Fanconi Anemia Endocrine Abnormalities

Case report

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Introdution:

Fanconi anemia (FA) is a rare disease, genetically and phenotypically heterogeneous, with recessive autosomal or X-linked transmission. It's a chromosome instability disorder, characterized by multiple congenital anomalies, bone marrow failure, and increased susceptibility to specific malignancies. Other findings, including short stature, skin pigmentation, and **endocrine abnormalitie**s have been recognized, most notably growth hormone deficiency (GHD), hypothyroidism, and hypogonadism.

This report includes **3 patients** with FA referred to pediatric endocrinology consultation at our Hospital. Patient 1 and 2 are siblings, children of consanguineous parents

Case report 1

Female, 21 years old, diagnosed at 8 years

Past medical history

- vesicoureteral reflux and neurogenic bladder
- encephalocele (surgery at 29 days old)
- conductive hearing loss
- strabismus, myopia and astigmatism;
- skeletal malformations (blocK vertebra C2-C4, vertebral dysmorphia)

12 years → • menarche

13,5 years → • dyslipidemia

18 years → • Metformin + simvastatin

12 years old
Impaired glucose tolerance

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- **without GHD**
- **regular height velocity**

PHYSICAL EXAMINATION

- "café-au-lait" spots (torso and limbs)
- bilateral hearing aids and eyeglasses
- left thumb agenesis and right thumb hypoplasia
- agenesis of the thenar muscles
- short stature (P3)
- overweight (P85-95)

Final adult height 151.5 cm

Case report 2

Male, 11 years old, neonatal diagnosis

Past medical history

- horseshoe kidney
- left inguinal hernia
- conductive hearing loss
- astigmatism

GHD → • clonidine and insulin hypoglycemia tests

MRI → • small pituitary gland

9 years old → • Somatropin

7 years 10 months old
Short stature

↓

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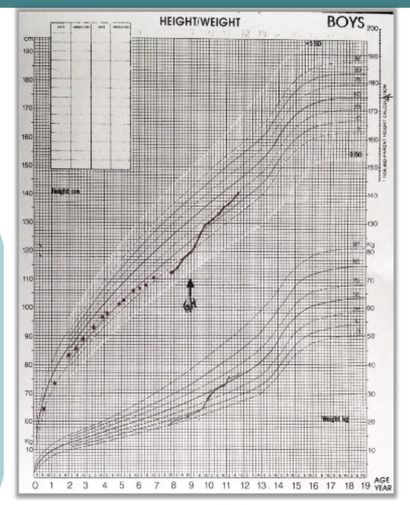
PHYSICAL EXAMINATION

- "café-au-lait" spots
- eyeglasses
- thumb hypoplasia and assymetria
- phimosis and left cryptorchidism
- weight -1.55 SDS
- height -2.16 SDS
- height velocity 1.68 SDS

Currently:
somatropin 27 ucg/Kg/day
weight -0.15 SDS
height -0.75 SDS
height velocity 6 cm/year
prepubertal



Tanner-Whitehouse curves

Case report 3

Male, 5 years old, diagnosis at 3 years and 7 months

Past medical history

- premature of 35 weeks
- fetal growth restriction
- Hydrocephalus and corpus callosum hypogenesis
- intermittent exotropia, myopia, astigmatism

Endocrine evaluation → • without GHD, normal lipidic profile and tiroid function

MRI → • corpus callosum hypogenesis

13,5 months old
Short stature

↓

Pediatric Endocrinology Unit

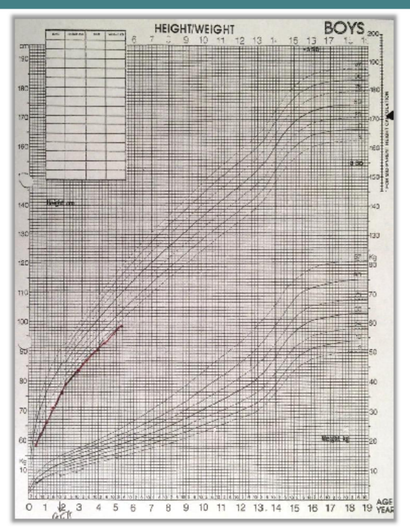
- **bone marrow failure**

Allogeneic transplantation
March 2015

PHYSICAL EXAMINATION

- "café-au-lait" spots
- short and narrow palpebral fissures
- short philtrum
- weight -3.49 SDS
- height -4.02 SDS

Currently :
weight -3.43 SDS
height -2.49 SDS
height velocity -0.46 SDS



Tanner-Whitehouse curves

Conclusion:

We pretend to emphasize the importance of periodic endocrine evaluation for patients with FA, looking for precocious diagnosis and treatment, Knowing that low number of cases and phenotypic diversity, make difficult follow-up.

In the particular case of GH treatment in FA patients, long-term risk is unknown, therefore, continued surveillance is needed, considering the increased risk for solid tumors in FA patients. We question the relevance of treatment with somatropin in FA patients without GH deficiency.

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Miscellaneous
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