**Fanconi Anemia Endocrine Abnormalities**

**Case report**

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**Introduction:**

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 abnormalities, 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
  - encephalcele (surgery at 29 days old)
  - conductive hearing loss
  - strabismus, myopia and astigmatism;
  - skeletal malformations (block vertebra C2-C4, vertebral dysmorphia)

- **12 years old:**
  - Impaired glucose tolerance
  - Pediatric Endocrinology Unit
  - Metformin + simvastatin
  - without GHD
  - regular height velocity

- **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

- **7 years 10 months old:**
  - Short stature
  - Pediatric Endocrinology Unit
  - GHD
  - clonidine and insulin hypoglycemia tests
  - MRI: small pituitary gland
  - Somatropin
  - 9 years old

- **Currently:**
  - GHD
  - Metformin + simvastatin

### 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

- **13.5 months old:**
  - Short stature
  - Pediatric Endocrinology Unit
  - MRI: without GHD, normal lipidic profile and thyroid function
  - corpus callosum hypogenesis

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

### 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.