ENDOCRINOLOGICAL ASSESSMENT IN PATIENTS WITH FANCONI ANEMIA

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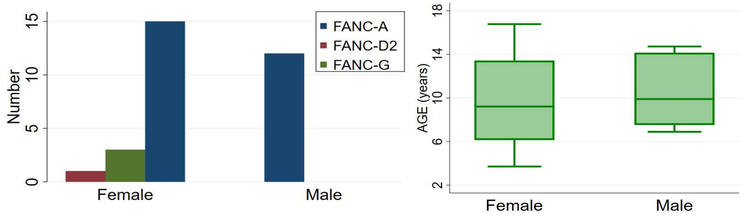
INTRODUCTION: Fanconi anemia (FA) is a rare genetic disease that presents with aplastic anemia. Around 60% have short stature (SST), with a mean height of -2.2 SD. However, studies assessing the etiology of SST not having reached relevant conclusions.

AIM: To Evaluate the clinical features and endocrine status in relationship to SST in patients with FA.

METHODS AND PATIENTS: A cross-sectional study was carried-out between 2019-2020 in **31 pediatric patients (19 females)** with FA. Auxological assessment, biochemical analysis, study of the hypothalamic-pituitary axis and an MRI of the hypothalamic-pituitary area were performed

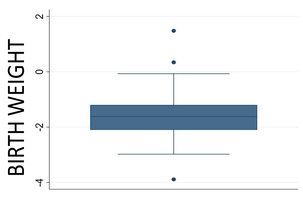
RESULTS:

PATIENTS

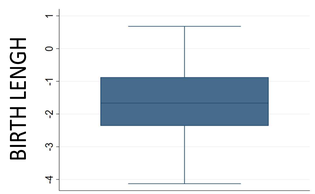


NEWBON AUXOLOGY

- ✓4 were NBPT
- ✓9 out of 26 had BW < -2 SD
- ✓10 ouf of 26 had BL <-2 SD



BW: -1.62 SD (*IQR*:-2.1; -1.2)



BL : -1.67 SD (*IQR*:-2.36; -0.88)

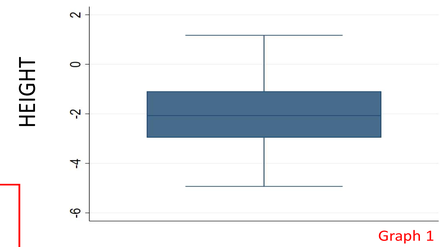
No correlation was found between height and anthropometry at birth

ENDOCRINOLOGICAL EVALUATION

The mean age at first evaluation was 9.7 ± 4.2 years : Tanner I (n=21), Tanner II (n=1), Tanner III (n=1), Tanner IV (n=2), Tanner V (n=6)

- The height was below -2 SD in 54.8% (17/31) Graph 1
- The mean BMI was -0.74 SD (± 0.83)
- 86% (19/22) being shorter than their target height (TH)
- Six female patients reached final height and were -2.3 SD (± 0.82) from their TH.
- Hematopoietic cell transplant (HCT) was performed in 16 patients.
- The attributable fraction of exposure to HCT in patients with SST was 42% [CI95% (71-85)], p>0.05.

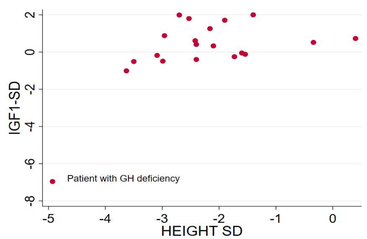
Only 5 patients had endocrinological alterations:
1= growth hormone (GH) deficiency
2= adrenal insufficiency secondary to corticosteroid treatment
2= had hypergonadotropic hypogonadism after transplantation



Graph 1

Height 2.07 SD (*IQR*: -2.96, -1.09)

GH/IGF AXIS



- ✓ Serum levels of IGF-1 and IGFBP-3 were normal in all patients, with the exception of GH-deficient patient.
- ✓ Staniocalcin 1 and 2 levels were lower in pubertal patients compared to healthy controls.

MRI FINDINGS:

- The volume of the pituitary gland was normal [0.45SD (IQR: -0.21; 0.75)], except in the patient with GH deficiency, who had a volume of -2.98 SD with ectopic neurohypophysis.

CONCLUSIONS:
Short stature is an integral feature of FA but does not seem to be related to a GH deficiency. However, an alteration in the peripheral regulation of the GH/IGF axis cannot be excluded.

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Presented at: ESPE 2021
Growth and syndromes (to include Turner syndrome)
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