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:

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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