Characterization of puberty development in a large cohort of patients with Noonan syndrome with molecular diagnosis

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**INTRODUCTION**

Noonan Syndrome (NS) is a relatively frequent multisystemic genetic disorder, characterized by typical facial abnormalities, congenital heart disease and short stature [1]. Despite the description of pubertal delay in both sexes, its pathophysiological explanation is still uncertain. The present study aims at characterizing pubertal development in NS and identifying predictors of pubertal delay.

**METHODS**

We analyzed 116 individuals with molecular diagnosis of NS and puberty evaluation. We characterized delayed puberty as pubertal onset after 12 years in girls and 13.5 years in boys, according to parameters of the Brazilian population [2,3]. To investigate pubertal delay predictors, we divided the sample into two groups according to the presence of normal or delayed puberty and correlated them with clinical characteristics.

**RESULTS**

The mean age at puberty for girls was 11.9±1.9 years and for boys, 12.5±1.7 years, significantly later than the Brazilian population (p = 0.025; p <0.001) (Figures 1 and 2). We identified pubertal delay in 27 (49.1%) girls and 17 (27.9%) boys (p = 0.031). Height gain from onset of puberty to adult height was lower in children with pubertal delay. The other differences for both sexes between the pubertal delay and normal puberty groups are described in table 1.

**CONCLUSION**

Pubertal delay is characteristically found in children with NS, more frequently in females. The low BMI of girls with NS could modulate the age of puberty, just as the increase in overweight/obesity in the general population has shown an effect on reducing the age of onset of puberty for girls [4].

**REFERENCE S**


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**Table 1 - Comparison of clinical characteristics between delayed and normal puberty groups in both sexes**