NOONAN SYNDROME PATIENTS WITH SHORT STATURE AT A SINGLE PEDIATRIC ENDOCRINOLOGY CENTRE

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INTRODUCTION

Noonan syndrome (NS) is caused by mutations in RAS/MAPK signaling pathway genes.

About 70% of the NS patients have short stature, and recombinant human growth hormone (rhGH) is an established yet not fully standardized treatment.

AIM

To assess the first 2 years of rhGH treatment effectiveness in NS patients at a single centre

METHOD

- A total of 20 (16 male) NS patients, diagnosed based on the Van der Burgt et al. criteria[1]
- 7 patients were treated with rhGH of whom 6 had at least 2 years of follow-up and were included in the analysis.
- Patients underwent anthropometry, clinical and laboratory investigations 6-monthly, echocardiography and bone age estimation yearly.

RESULTS

- DNA test results of the treated patients are presented on Fig. 1
- Mean age at NS diagnosis - ***7.8±3.4 years*** (1.3÷10.5),
- Mean age at rhGH start - ***9.1±1.5 years*** (7.5÷10.7).
- Treatment period - ***38.3±15.3 months***
- Baseline SDSheight ***–3.42±0.58*** (-4.1 ÷ -2.6), SDSweight ***–3.07±0.58*** (-3.73 ÷ -2.27), SDS_{IGF1} ***–1.12±0.98*** (-2.44÷ 0.25)
- BA delay at diagnosis was ***2.6±0.9 y.***
- RhGH starting dose ***0.035±0.005mg/kg/d***, slightly increasing by the end of the 1st year (0.036±0.002 mg/kg/d), and 2nd year (0.037±0.003 mg/kg/d).
- The 1st and 2nd year growth velocity is presented on Fig. 2
- The 1st year ΔSDSheight was ***0.72*** (p=0.002), ΔSDSweight was ***0.83*** (p=0.025). The 2nd year ΔSDSheight, ΔSDSweight and ΔSDSBMI increased insignificantly. (Fig. 3 and Fig. 4).
- ΔSDS_{IGF1} were ***1.70*** (p=0.067) and ***0.25*** (n.s.), respectively.
- By the end of the 2nd year, the mean BA remained significantly delayed.
- No treatment side effects were observed

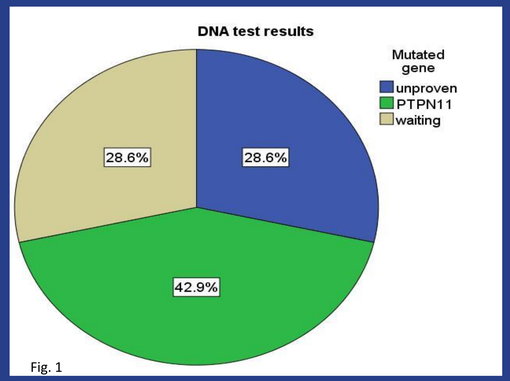

Fig. 1

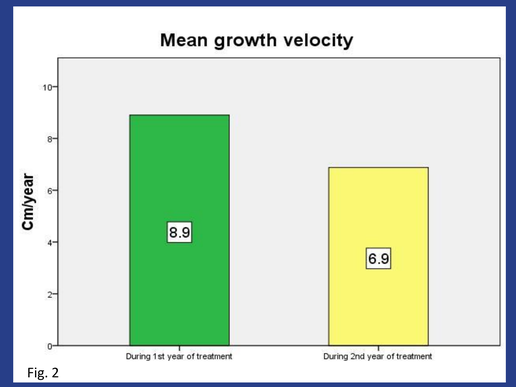

Fig. 2

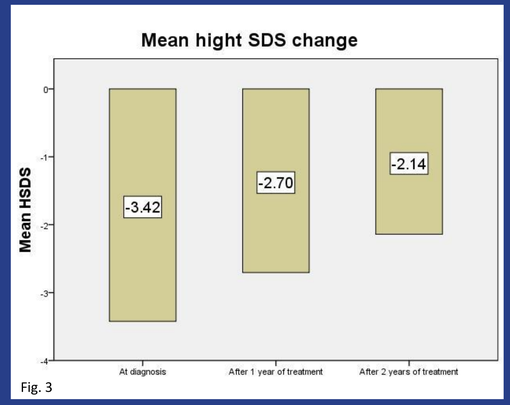

Fig. 3

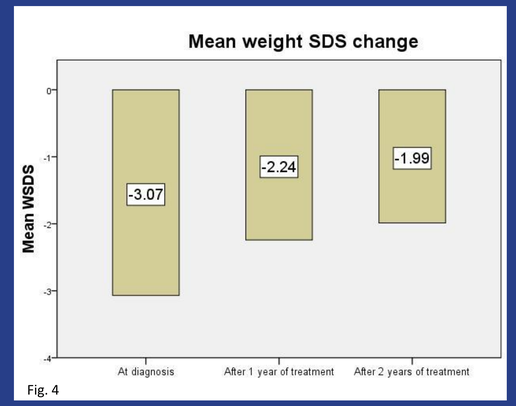

Fig. 4

CONCLUSIONS

Our study showed that NS patients follow the general patterns for the first 2 years of rhGH treatment.

The applied doses seem insufficient to cause good height increment.

In order to improve outcomes, the treatment should be further standardized.

REFERENCES

1. Van der Burgt I, Berends E, Lommen E, van Beersum S, Hamel B, Mariman E. Clinical and molecular studies in a large Dutch family with Noonan syndrome. Am J Med Genet. 1994 Nov 1;53(2):187-91. doi: 10.1002/ajmg.1320530213. PMID: 7856646.

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