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.

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

- 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.

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


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**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±2.58 (-4.1± -2.6), SDSweight -3.07±0.58 (-3.73 ± -2.27), SDSOF -1.12±0.98 (-2.44± 0.25)
- BA delay at diagnosis was 2.6±0.9 y.
- RhGH starting dose 0.035±0.005 mg/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).
- ΔSDSOF 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.

**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.