**INTRODUCTION**

- Besides an excessive, early-childhood weight gain, hyperphagia is the key symptom in patients with monogenic obesity [1, 2].
- However, the assessment of hyperphagia is still challenging [3].

**AIM**

Implementation of the hyperphagia questionnaire developed for patients with Prader-Willi Syndrome (PWS) in patients with monogenic obesity to assess the severity of hyperphagia.

**METHODS**

- Enrollment of pediatric patients with biallelic pathogenic leptin receptor (LEPR) variants, heterozygous pathogenic melanocortin-4 receptor (MC4R) variants and 16p11.2 microdeletion including deletion of Src homology 2B1 (SH2B1)
- Assessment of the 13-item hyperphagia questionnaire from Dykens et al. [4] by their parents, developed and validated to assess hyperphagia in patients with PWS
- Items were summarized in a total hyperphagia score and its subscores hyperphagic behaviour, hyperphagic drive and hyperphagic severity

**RESULTS**

- Enrollment of 20 children with monogenic obesity (Table 1)
- Significant differences in BMI z-score, body fat ratio, total leptin and bioactive leptin levels between patients with LEPR variants and MC4R variants or 16p11.2 deletions (p < 0.05; Table 1)
- Significantly higher total hyperphagia scores in patients with LEPR and MC4R variants compared to patients with 16p11.2 deletions (p < 0.05, Figure 1)
- Moderate correlation between the age of all patients and the scores total hyperphagia (r = -0.456, p < 0.05) and hyperphagic behavior (r = -0.516, p < 0.05)

**CONCLUSIONS**

- Hyperphagia scores are comparable in patients with LEPR and MC4R variants
- Patients with 16p11.2 deletions show less severe hyperphagia scores than patients with LEPR or MC4R variants
- Inverse relationship between age and total hyperphagia score as well as hyperphagic behaviour subscore suggests that the severity of hyperphagia declines with age
- Dykens’ hyperphagia questionnaire is a useful tool to assess the severity of hyperphagia in patients with monogenic obesity

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**REFERENCES**


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**Figure 1:** Total hyperphagia score and hyperphagic behaviour subscores in patients with leptin receptor (LEPR) variants, melanocortin-4 receptor (MC4R) variants and 16p11.2 deletions. Data are presented as median and interquartile range. Significant differences between groups were analysed using Kruskal-Wallis test. Mean values ± (SD) for individuals with severe obesity without a genetic cause published by Sherer et al. [5] are marked by grey line and grey area. Dotted lines indicate the maximum and minimum scores that could be reached.