

QUALITY OF LIFE IN CHILDREN WITH ACHONDROPLASIA AND PARENTAL WELL-BEING EVALUATION

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INTRODUCTION

Achondroplasia is an autosomal dominant genetic disease caused by a mutation in the Fibroblast Growth Factor Receptor 3 (FGFR3) gene. The condition is characterized as an epiphyseal dysplasia, manifesting as a disproportionately short stature. In addition to the comorbidities and dysmorphologies associated with this diagnosis, the impact of achondroplasia on patients' quality of life, as well as that of their parents and caregivers, is well recognized.

OBJECTIVES

To evaluate the quality of life in children with achondroplasia and the well-being of their parents.

METHODS

This is a retrospective observational study using the Achondroplasia Child Experience Measures (ACEMs) and the Achondroplasia Parent Experience Measure (APEM) questionnaires. The questionnaires were structured using a Likert scale (ranging from 1 to 5, with higher numbers indicating a greater impact on quality of life). The data are presented as mean $\pm$ standard deviation (SD), calculated using the 'R for Windows' software.

RESULTS AND DISCUSSION

The sample consisted of 12 patients with achondroplasia (6/12 male), with a mean age of 9.41 ± 3.62 years, and one parent of each child ($n = 12$). The average scores for the domains assessed in the ACEMs questionnaire were as follows: Physical symptoms/complications – 1.60 ± 0.40 , Functioning – 1.96 ± 0.70 , Emotional well-being – 1.85 ± 1.28 , Social well-being – 2.11 ± 0.76 , and Need for assistance/adaptative services – 3.03 ± 0.82 points. Regarding the APEM questionnaire, the mean scores were: Caretaking responsibilities – 3.24 ± 0.90 , Emotional well-being – 3.44 ± 1.28 , Family – 1.41 ± 1.16 , and Work – 2.16 ± 1.52 points (figure 1).

This study assessed the quality of life of children with achondroplasia, revealing a greater impact on the need for adaptive assistance and services, as well as on social well-being. In this regard, it has been established that the biomechanical and anatomical alterations characteristic of achondroplasia are associated with delays in psychomotor development and increased reliance on external support. For parents, there was a greater impact on caregiving responsibilities and emotional well-being, with minimal impact on family life or work-related activities. It is important to note that the questions refer to the past two weeks, so scores may vary significantly depending on the timing of the survey.

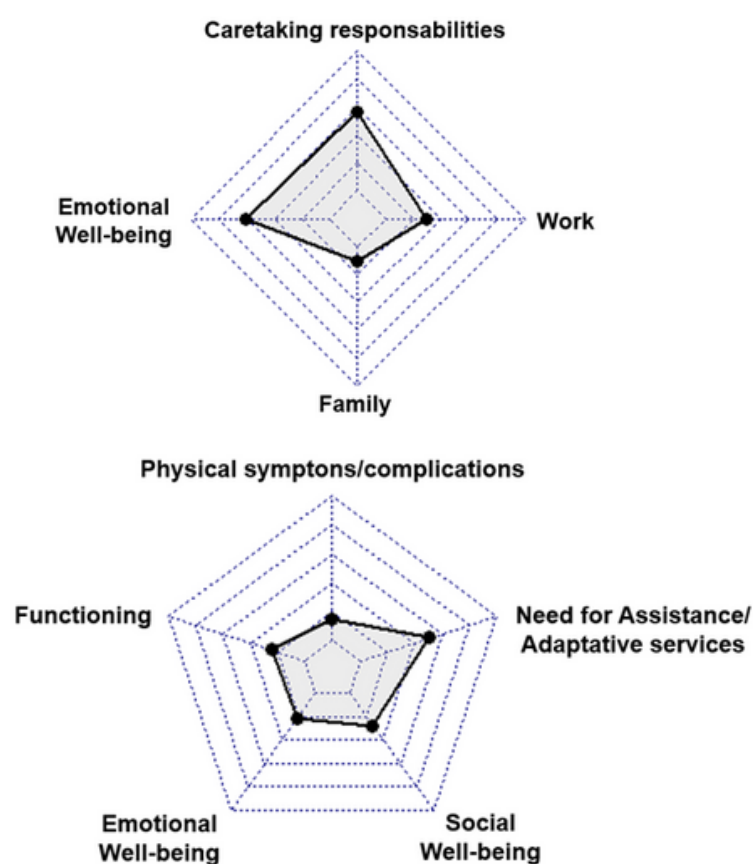


Figure 1: Distribution of mean scores for the domains assessed in the ACEMs and APEM questionnaires.

CONCLUSIONS

The psychological well-being of both patients and their parents or caregivers should be regularly assessed, and the use of the ACEM and APEM questionnaires may represent suitable tools for children aged between 2 and 12 years, as they address multiple dimensions of life. Referral for psychological counseling may be necessary.

REFERENCES:



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