GROWTH AND PUBERTY IN PATIENTS WITH OSTEONECOSIS IMPERFECTA

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INTRODUCTION

- Osteogenesis imperfecta (OI) is a heritable systemic connective tissue disorder characterized by low bone mass, bone fragility, impaired linear growth, short stature, and bone deformities.
- Patients with OI are classified by the Sillence classification that designates patients based on clinical features and severity of the disease.

AIM

The study aims to evaluate growth and pubertal characteristics of 83 patients with OI.

METHOD

- This study was designed as a retrospective study involving children with OI who received care at the Pediatric Endocrinology Department of Istanbul University Faculty of Medicine.
- Medical charts of patients who were followed up every 3-6 months between 1992-2019 were evaluated.
- Demographic findings, data on birth status (gestational age, birth weight, and length), clinical features, growth and pubertal data, and laboratory findings were gathered from medical files.

RESULTS

- 83 (31 female/52 male) patients were enrolled.
- The median follow-up duration was 4.7 (0.6-17.7) years.
- 51 out of 83 patients (61.4%) received bisphosphonate therapy.
- The median Z-score of the bone mineral density improved in patients with OI-I and OI-III with the treatment.
- During follow-up, height-SDS significantly increased in both OI-I and OI-III; however, final adult height SDS of patients did not improve.
- The frequency of overweight and obesity was found to be increased at the last evaluation compared to the admission.
- The rate of precocious puberty (PP) and early puberty (EP) were 20% and 10% in girls, and they were 15.7% and 47.3% in boys, respectively.

CONCLUSIONS

- FAH-SDS did not improve despite remarkable amelioration in BMD-2 scores on therapy.
- There is a trend towards earlier puberty in OI patients, and the frequency of overweight and obesity was found to be increased over time.
- Reduced growth, significant weight gain over time due to impaired mobility, and high frequency of PP/EP require effective interventions to improve mobility and functional parameters as early as possible in children with OI.

REFERENCES


ACKNOWLEDGEMENTS

We thank all patients and their parents for participating in this study.

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