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GROWTH IN OSTEOGENESIS IMPERFECTA

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INTRODUCTION

Osteogenesis imperfecta (OI), or brittle bone disease, is a rare disorder with congenital bone fragility caused by mutations in the genes that codify for type I pro-collagen production in osteoblasts (*COL1A1* and *COL1A2*), located in chromosomes 7 and 17.¹ Numerous mutations have been described as causing the condition.² In the vast majority of cases, OI is inherited in a dominant fashion, or caused by a new mutation. The prevalence of OI is estimated to be 1 in 20 000 to 50 000 infants.³

Besides brittle bones, clinical characteristics and severity of OI are widely variable. There

may even be a different degree of severity in different members of the same family.^{4,5} Clinical features that may be present include bone fragility, joint hyperlaxity, muscle weakness, chronic unremitting bone pain, and skull deformities (eg, posterior flattening) due to bone fragility in infants with severe OI. Fractures may still occur after puberty,⁶ with bone fragility persisting throughout life. Individuals with mild forms of the disease may have normal stature with no deformities or fractures at all, and the condition would be diagnosed only when an x-ray is obtained for other reasons. People with severe OI may have extreme short stature and severe deformity of the long bones. Exercise tolerance and muscle strength are significantly reduced in patients with OI, even in the mild forms.⁷

Osteogenesis imperfecta can affect several organs and systems. For example, hearing loss may be present in about 50% of the

From The Editor's Desk

Dear Colleague:

You may be aware that our former sponsor, Insmed, settled a patent infringement dispute and no longer promotes IGF-I/IGFBP-3 to patients with severe primary IGF-I deficiency or other short stature indications. Therefore, they no longer provide an educational grant to the GGH journal. Consequently, Pediatric Sunshine Academics, Inc., a 501(c)(3) non-profit organization, is funding the cost of this issue of GGH without prior anticipation or alternative funding sources available.

However, I am committed to seek new grants that will allow us to continue publishing this journal. I am grateful to the editorial board for their strong support; they have all pledged to contribute with their usual efforts and expertise while we seek more stable times. Since its inception 23 years ago, GGH has improved and expanded; it is held in high regard and enjoys over 11 000 subscribers. We all feel obliged not to let you down.

In order to forge ahead GGH will need the support of its readers while we elicit educational grants. You can help us during this transition by contributing to Pediatric Sunshine Academics, Inc. an organization whose mission is to support research and education in pediatric endocrinology and nutrition. Your fully tax deductible donation to **Pediatric Sunshine Academics, Inc., P. O. Box 3208, Tallahassee, FL 32315-3208**, either by check or online at www.PedSacademics.org will be used entirely for the continued publication of GGH. Pediatric Sunshine Academics, Inc.'s federal EIN is 65-0854085.

On behalf of the editorial board, I thank you in advance for your donations and support. I will keep you apprised of our quest to elicit new grants and sponsorships for the continuation of the publication of GGH.

Fima Lifshitz, MD
Editor-in-Chief