

CLINICAL FEATURES IN SHOX HAPLOINSUFFICIENCY: DIAGNOSTIC AND THERAPEUTIC IMPLICATIONS

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INTRODUCTION

The distal end of Xp and Yp is composed of 2.6 Mb DNA sequences that are identical between the X and the Y chromosome.¹ This particular region is named the short arm pseudoautosomal region (PAR1), where the X and the Y chromosomes recombine during male meiosis.¹ Since Xp terminal deletions invariably result in short stature irrespective of the breakpoints,² and small Yp terminal deletions lead to short stature,³ it has been suggested that a growth gene escaping X-inactivation resides in the PAR1, and that haploinsufficiency of the growth gene causes short stature in both sexes as a dominant phenotype.²

In 1997, Rao et al⁴ successfully cloned a novel gene at the position roughly 500 kb from the Xp/Yp telomere, and named it SHOX for short stature homeobox containing gene. SHOX consists of 7 exons and produces 2 transcripts

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From The Editor's Desk

The miracle of the Internet has allowed the readership of *Growth, Genetics & Hormones* to grow very rapidly. We have recently added to our subscribers a substantial number of pediatric endocrinologists worldwide. Members of the Pediatric Endocrine Societies from Europe, Latin America, Colombia, and Japan who have email addresses will now be receiving *GGH* on an ongoing basis. It gives me great pleasure to welcome these pediatric endocrinologists to the family of *GGH*. Surely, our European, Latin American & Japanese contemporaries will help us broaden our perspectives and apprise us of advances in the field for publication in *GGH*. I am looking forward to contributions from our colleagues; an example of such is the lead article in this issue.

This second issue of 2004 contains a review of the clinical features of the short stature homeobox gene, so called SHOX. This important factor is implicated in the etiology of short stature and, in particular, features that characterize patients with this abnormality. This paper addresses a complicated subject, presents it in a clear easy-to-read manner, and brings the state of the art in the field to the readers of *GGH*. Drs. Tsutomu Ogata and Maki Fukami from Tokyo, Japan authored this lead article, emphasizing aspects of particular interest to pediatric endocrinologists and geneticists. The authors deserve our congratulations and thanks for their erudite writing.

This issue also contains abstracts of recent articles published in the literature that were considered of importance by our editorial board; each article is reviewed with editorial comments. Unfortunately, we have limited space and cannot publish all articles of importance in the field, nor do we attempt to do so. We limit our scope to bring value by publishing only articles that attract the interest of the editorial board and that meet our editorial standards. The high value that *GGH* has received from the readership indicates we have met our objectives, and we want to surpass them. The report of the December 2003 survey is posted at www.GGHjournal.com (click on survey results). We appreciate your comments so we may continue to serve your needs.

Fima Lifshitz, MD
Editor-in-Chief

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