

GROWTH AND GROWTH HORMONE IN CHILDREN WITH NEUROFIBROMATOSIS TYPE 1

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INTRODUCTION

Neurofibromatosis type 1 (NF1), also known as von Recklinghausen disease, is an autosomal dominant, commonly inherited disease that affects one of every 3000 individuals.¹ The gene responsible for this condition has been isolated by positional cloning to chromosomal region 17q11.2. It spans over 350 kb of genomic DNA and encodes neurofibromin, a protein product of 2818 amino acids that is expressed in various tissues.² According to the National Institutes of Health Consensus Development Conference (Bethesda, Maryland, July 13-15, 1987), there are 7 key components of the disease (Table 1), at least 2 of which must be present in order to establish the diagnosis.³

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From The Editor's Desk

In this issue the international contributing editors of *GGH* began abstracting papers for the readership. They reviewed the literature, selected important publications, abstracted them, and made insightful and important comments. Thus, with these contributions we continue to expand the reach of the journal. On behalf of everyone I thank them for their contributions and welcome them.

The lead article covers an important area that clinicians often encounter, namely evaluating the growth of patients with neurofibromatosis type 1. Growth alterations affect 13% to 24% of children with this disease, with more than 40% of adult NF1 patients reaching a decreased final height. Drs. Karantza and Geffner have written a notable review outlining the most pertinent issues of growth and growth hormone in NF1 patients. I am sure that this lead article will be tremendously useful and will constitute an important reference.

Additionally, the highlights of the 87th annual meeting of the Endocrine Society are summarized and 8 abstracts are presented in this printed *GGH*. There are 7 more abstracts posted on the web at www.GGHjournal.com. I trust you will enjoy and treasure this issue.

The next issue will include a historical review of growth hormone that commemorates the 20th anniversary of the FDA approval of recombinant human growth hormone and the launching of *GGH*—both made possible by Genentech.

Fima Lifshitz, MD

BACKGROUND

Endocrine disorders have been reported in approximately 1% to 3% of all NF1 patients. Pheochromocytoma is the most common endocrinopathy in adults with NF1, occurring in approximately 1% of patients.⁴ In children with NF1, the most prevalent hormonal disorder is central precocious puberty (CPP), with a frequency of 3% compared to 0.06% in the

