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SF1 MUTATION IN HUMANS

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INTRODUCTION

The purpose of this lead article is to bring readers up to date on the phenotypes, genotypes, and pathogenesis of the steroidogenic factor (SF)1 mutation that pediatric endocrinologists encounter in their practices and to provide new insights into SF1 function in humans. Steroidogenic factor 1 (Sf1 in mice or SF1 in humans), also called Ad4BP or NR5A1, is a nuclear transcriptional factor that binds to target gene promoters as a monomer and recognizes a canonical half-site motif. Structurally, both Sf1 and SF1 have characteristic domains

of nuclear transcriptional factors. These consist of a zinc finger DNA-binding domain, a ligand-binding domain, and an activation function-2 domain. There is also an accessory DNA-binding domain that confers binding site stability and specificity.

Originally, SF1 was isolated as a global regulator for P450 steroid hydroxylases.^{1,2} SF1 was thought to be responsible for tissue-specific expression of these enzymes in the adrenals and gonads. Subsequent studies in vitro have shown that Sf1 and SF1 regulate a lot of genes involved in adrenal and gonadal development, sex differentiation, steroidogenesis, reproduction, and many other metabolic functions.^{2,3} Thus, Sf1 and SF1 play pivotal roles in the development and function of multiple endocrine organs.

From The Editor's Desk

This issue of *GGH* Volume 24, Number 1 is only available on-line and will be not be printed and mailed due to budgetary constraints. However this issue is available either as a PDF file or a web page so you can file it and/or print it and keep it for your enjoyment and as a reference resource.

The current issue includes an excellent and timely review of the "SF1 Mutations in Humans" by Dr. Tomonobu Hasegawa, plus 19 reviews of current papers in the literature with comments by the editorial board. There are four reviews pertaining to growth hormone treatment including the consensus guidelines of adult growth hormone deficiency, two addressing growth of celiac patients, three pertaining to height related issues on quality of life, the in vitro fertilization children or the genetics of stature. There are also two reviews regarding the aortic dilatation and the uterine development of Turner patients. In addition the late effects on cancer survivors, hypopituitarism following traumatic brain injury, and diabetes and stroke in hypopituitarism are also reviewed. I also want to bring to your attention the reviews on the FTO gene in obesity and the monoallelic expression of autosomal genes. Finally there are two reviews of papers dealing with two frequent alterations in pediatric endocrine practices, namely metabolic syndrome in brothers of PCOS women and the ventricular function of congenital hypothyroidism in neonates.

The economic situation in the country is being reflected in our journal. The reduced funding for continuous medical education will only allow us to publish two electronic issues in 2008, unless there is a renewed commitment for sponsorships that will allow us to provide our readers with a high quality journal more frequently. We will continue to search for means and will appreciate your tax deductible contributions. You may do so on line (www.GGHjournal.com or PedsAcademics.org) and click *make a donation*, or you may send a check to Pediatric Sunshine Academics, 1040 Alston Rd., Santa Barbara, CA 93108.

Thank you for your support,
 Fima Lifshitz, MD
 Editor-in-Chief