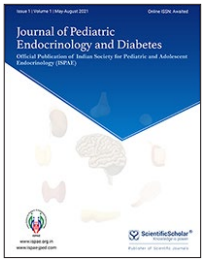
ScientificScholar®
Knowledge is power
Publisher of Scientific Journals

https://ispae-jped.com/

Journal of Pediatric Endocrinology and Diabetes



Editorial

Editor's page

P. S. N. Menon[1]

[1]Former Professor of Pediatrics and Consultant-in-Charge Pediatric Endocrinology, All India Institute of Medical Sciences, New Delhi, India.

***Corresponding author:**
P. S. N. Menon,
Former Professor of Pediatrics and Consultant-in-Charge Pediatric Endocrinology, All India Institute of Medical Sciences, New Delhi, India.

psnmenon@hotmail.com

Received: 24 July 2024
Accepted: 24 July 2024
Published: 10 August 2024

DOI
10.25259/JPED_31_2024

Quick Response Code:

We welcome you all to another interesting issue of our journal, covering a collection of original articles, case reports, images, reviews, and journal updates. We do hope these articles are academically exciting to the readers, especially the postgraduate students and fellows in training.

Idiopathic short stature (ISS) is defined as a height more than two standard deviations below the corresponding mean height for age, sex, and population in a child with normal birth size and normal body proportions and without evidence of any systemic, endocrine, nutritional, or chromosomal abnormalities. It is often a diagnosis of exclusion. Growth hormone (GH) is most commonly tried in the treatment of ISS and is recommended for use in children with ISS in some of the countries. Third-generation aromatase inhibitors (AIs) such as letrozole or anastrozole have been shown to effectively suppress estrogen biosynthesis and delay bone maturation in children with ISS, resulting in an increased predicted adult height following 1–3 years of treatment. In this issue, Dutta *et al.* from Delhi and Mumbai, India, describe the results of a meta-analysis done to evaluate the efficacy and safety of third-generation AIs in the management of ISS. Although the number of studies is few, there are some reassuring data on the efficacy and tolerability of AIs on height outcomes and pubertal progression outcomes in ISS. The authors opine that there is a scope for using AIs and GH together in children with ISS to have a synergistic impact on height outcomes. However, dedicated long-term studies are needed in this area.

In an invited editorial commentary on this topic, Atlas and Zacharin from Melbourne, Australia, note that the meta-analysis addresses a management strategy to increase adult height in the case of normal variant short stature or ISS. An important question to be addressed is that of the intrinsic value and social significance of seeking greater height as a desirable commodity. The authors also question the methodologies that may be utilized in attempting to achieve these outcomes. There are growing concerns about the use of pharmacological interventions (including GH) aiming to augment height beyond a child's genetic potential. The recommendation for the use of AIs calls for more evidence on safety and efficacy before routine prescriptions for children with short stature could be supported. The authors express their concern about extending the rationale for the use of AIs in specific cases to include ISS.

Simon from Vellore, India, discusses the Indian perspective on delaying the growth plate closure to augment height. There are many clinical observations that adult height is dependent on the timing, duration, and level of circulating estrogens. One of the reasons for not advising the use of AIs in prepubertal boys is due to an association with vertebral deformities. Moreover, children with ISS represent a highly heterogeneous group with multiple differing extrinsic and intrinsic growth plate defects, and as such, the response to hormonal or pharmacological intervention may be variable. She also reminds readers that the use of anti-estrogens in the management of short stature is only acceptable in pubertal boys now. Caution is the catchphrase in such management strategies.

We have recently published several articles on various aspects of diagnosis and optimal management of congenital hyperinsulinemia of infancy (CHI). In the section on "Genetics for the Pediatric Endocrinologist," Mittal *et al*. from New Delhi, India, review the molecular mechanisms underlying CHI and their relevance to management. The majority of the cases relate to defects in K-ATP channels that regulate insulin secretion from pancreatic beta-cells. These are mostly attributable to mutations in *ABCC8 and KCNJ11*, both located on the short arm of chromosome 11, that code subunits of the K-ATP channel (SUR and Kir6.2, respectively). However, the underlying molecular defect may be identified in only half of them. A critical sample drawn at the time of hypoglycemia is crucial for biochemical characterization and is the beginning of a cascade of investigations that further elucidate further course of action.

This issue also has some unusual, yet noteworthy case reports.

Siriwardhane *et al*. from Colombo, Sri Lanka, describe a case series with clinical characteristics and management of gonadotropin-independent precocious puberty in four children with McCune–Albright syndrome (MAS) from a tertiary care center. Management options are varied with AIs showing promising results. It is of utmost importance to follow-up children with MAS regularly for the development of other hyperfunctioning endocrinopathies with regular biochemical assessment.

Mohan *et al*. from Calicut, India, describe a 3-year-old child with lower limb abnormalities and short stature with a similar history and findings noted in the father. The phosphate-regulating endopeptidase homolog X-linked gene variation was etiology. The authors highlight the importance of prompt genetic diagnosis and initiation of treatment to prevent subsequent sequelae.

Reddy and Bhattacharyya from Bengaluru, India, describe the case of a 9.5–year–old male child who presented with hyperpigmentation and normal male external genitalia. He was diagnosed with primary adrenal insufficiency, and whole exome sequencing revealed a homozygous mutation in the *STAR* gene consistent with lipoid congenital adrenal hyperplasia (LCAH), clinically fitting into the non-classical category corresponding to <10–20% residual enzymatic activity.

In the Images section, Marakkar *et al*. from Calicut present the images of a 16-year-old girl with Prader–Willi syndrome (PWS) with primary amenorrhea who had normal breast development and pubic hair growth from 11 years of age. Hypogonadotropic hypogonadism in PWS is not uncommon. The authors highlight that the diagnosis of PWS was missed during infancy and childhood. This oversight could have been prevented with greater awareness among treating clinicians regarding the clinical features and course of the PWS.

In our regular feature on "Ped Endo Journal Scan," Joshi, Brisbane, Australia, discusses five recent fascinating publications. These include evaluation of the ongoing trials of two new therapeutic agents – a phase 3 trial of crinecerfont in pediatric congenital adrenal hyperplasia and a phase 2, open-label, and multicenter trial of setmelanotide for the treatment of acquired hypothalamic obesity. Another interesting article is on the late endocrine effects of hematopoietic stem cell transplants in pediatric patients. A 25-year review on Klinefelter syndrome and high-grade aneuploidies will help to advance our knowledge on the expanding the gene-dosage effect of supernumerary X chromosomes. The study on clinical presentations and long-term outcomes in 144 patients with pediatric medullary thyroid carcinoma over six decades will definitely expand the horizons of management. The editors are of the opinion that these new research articles will add insights for better and optimal management of children with chronic endocrine disorders.

We have endeavored our best to present to you a variety of interesting clinical situations requiring astute observations, clinical acumen, and supportive laboratory in the diagnosis and management of common and not-so-common endocrine situations. We look forward to your comments and suggestions and welcome contributions to the forthcoming issues of our journal.

Happy reading!

How to cite this article: Menon PS. Editor's page. J Pediatr Endocrinol Diabetes. 2024;4:2-3. doi: 10.25259/JPED_31_2024