

Editor's Page

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P. S. N. Menon<sup>1</sup>

<sup>1</sup>Former Professor of Pediatrics, Divisions of Pediatric Endocrinology and Genetics, All India Institute of Medical Sciences, New Delhi, India.



**\*Corresponding author:**

P. S. N. Menon,  
Former Professor of Pediatrics,  
Divisions of Pediatric  
Endocrinology and Genetics,  
All India Institute of Medical  
Sciences, New Delhi, India.  
[psnmenon@hotmail.com](mailto:psnmenon@hotmail.com)

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We welcome you to yet another interesting issue of JPED, covering a selection of original articles, editorial commentaries, case reports, images, reviews, fellows' corner, and journal updates. We hope that these articles will intellectually stimulate readers, especially postgraduate students and fellows in training.

In our series titled "Invited review," Minu Bajpai, New Delhi, India, discusses the surgical aspects of genitoplasty for the management of 46,XX disorders of sex development (DSD) and congenital adrenal hyperplasia (CAH).<sup>[1]</sup> The issues discussed include gender assignment, timing of intervention, and long-term outcomes. Feminizing genitoplasty aims to restore functional anatomy, preserve urinary and sexual function, and support psychosocial wellbeing within a multidisciplinary and ethically informed framework. While many individuals with CAH retrospectively favor childhood surgery, population data show heterogeneous views, supporting a case-by-case approach rather than a universal moratorium. Ethical and legal standards emphasize transparent discussion of risks, alternatives, and long-term outcomes; consent must be qualified, persistent, written, and reaffirmed over time.

In a second "Invited review," Kanishka Das *et al.*, Bhubaneswar, India, discuss the various issues related to the surgical management of 46,XY DSD.<sup>[2]</sup> Collective insights into intermediate and long-term results of surgical cosmesis, functional outcomes, and gender dysphoria have tempered the surgical strategies with a global suspension on elective interventions till the age of consent. Besides the pros and cons of early versus late surgery for 46,XY DSD, the narrative also elaborates on the indications, surgical principles, timing of surgery, salient procedural details, and outcomes. Masculinizing procedures include gonadal surgery, penile reconstruction, surgery for urogenital sinus (UGS) and/or Müllerian remnants, and breast reduction. Likewise, feminizing procedures encompass clitorophallic reduction, surgery for UGS and/or vaginoplasty, gonadectomy, and breast augmentation.

Pilar Morillas-Amat *et al.*, Valencia, Spain, in an "Original article," evaluated the efficacy and safety profile of vosoritide treatment in children with achondroplasia after 12 months of administration retrospectively.<sup>[3]</sup> Changes in height Z-score, annualized growth velocity (AGV), arm span/height ratio, sitting height/standing height ratio, changes in bone age, and reported adverse events were analyzed. A significant increase in AGV was observed, along with an increase in AGV Z-score and height Z-score. Five patients did not show clinically relevant improvement in height Z-score. No serious adverse events were reported. The authors concluded that vosoritide treatment was generally effective and well-tolerated, leading to an average increase in height Z-score after 1 year, with no serious adverse effects. However, larger, long-term studies are needed to confirm these results and clarify the clinical impact.

In an accompanying "Editorial commentary," Shubha R Phadke, Pune, India, notes that the mechanism of action of vosoritide is targeted to the pathogenetic gain-of-function *FGFR3*

mutation causing achondroplasia and scientifically appears to be an effective way of treating the disorder.<sup>[4]</sup> When projected for the future, the estimated additional height gain for patients with achondroplasia treated with vosoritide versus those untreated was 20 cm for boys and 16 cm for girls over the 11 years from 6 to 16 years of age, with improvements in lumbar lordosis and knee curvature. The availability of targeted disease-modifying therapy brings a ray of hope to patients' lives.

In another "Original article," Orla Dempsey *et al.*, Glasgow, UK, describe a retrospective case review of newly diagnosed children with type 1 diabetes mellitus (T1DM) for thyroid and adrenal autoantibody status.<sup>[5]</sup> Overall, 14% developed hypothyroidism, with eight diagnosed at initial TPO antibody detection. Two developed hypothyroidism after 2 years. Adrenal dysfunction was infrequent, with only three children testing positive for adrenal antibodies and none developing adrenal dysfunction after a median of 4 years of follow-up.

In an accompanying "Editorial commentary," Anju Virmani, New Delhi, India, evaluates the implications of cost effectiveness of routine screening of thyroid, adrenal, and other autoantibodies in newly diagnosed T1DM children.<sup>[6]</sup> Current International Society for Pediatric and Adolescent Diabetes (ISPAD) guidelines state that screening children for thyroid antibodies can help stratify which youth with diabetes to follow most closely for development of hypothyroidism; however, in clinical practice, thyroid and adrenal antibody testing seems to fulfil very little purpose. The author strongly recommends a judicious choice of these tests in selected case scenarios.

We have three very interesting "Case reports" lined up for you. A salt-wasting crisis in neonates may arise from aldosterone deficiency, as seen in CAH, or from aldosterone resistance, as in pseudohypoaldosteronism. Differentiating between these two conditions is essential for appropriate management to optimize outcomes. VV Sivarama Raju *et al.*,<sup>[7]</sup> Hyderabad, India, report a 2-month-old male infant presenting with failure to thrive, vomiting, dehydration, and severe electrolyte abnormalities. Chemiluminescence assays may yield false-positive results for elevated cortisol and aldosterone levels due to cross-reactivity with steroid precursors. ACTH-stimulated 17-hydroxyprogesterone testing and genetic analysis are essential for accurate diagnosis and management.

Richa Arora *et al.*,<sup>[8]</sup> New Delhi, India, describe a 2-year-old boy with tall stature, diffuse hyperpigmentation, recurrent infections, and hypoglycemic seizures. Endocrine evaluation confirmed isolated cortisol deficiency with markedly elevated ACTH, and genetic testing identified a pathogenic *MRAP* variant. Hydrocortisone replacement led to improved pigmentation and stabilization of glucose.

Sitosterolemia is a rare autosomal recessive disorder of sterol metabolism, often misdiagnosed as familial hypercholesterolemia due to overlapping features of tendon xanthomas and severe hypercholesterolemia. Anukriti Agnihotry *et al.*,<sup>[9]</sup> Rishikesh, India, report a 1.5-year-old breastfed girl who presented with multiple tendon xanthomas, marked low-density lipoprotein-cholesterol elevation, and poor response to statins, dietary measures, and cholestyramine. Genetic analysis confirmed a compound heterozygous mutation in *ABCG8*. Following initiation of ezetimibe, her lipid profile normalized, and xanthomas stabilized without adverse effects.

In the "Images/Spotters" category, Sachin Kumar *et al.*,<sup>[10]</sup> Rishikesh, India describe reversible early-onset glycogenopathy (Mauriac syndrome) confirmed by liver histopathology in a child with T1DM. Daily insulin dose was subsequently titrated to achieve adequate glycemic control with complete resolution of hepatomegaly and transaminitis.

Under the series, "Fellows Corner," Lekshmi G,<sup>[11]</sup> Bengaluru, India, reflects how pediatric endocrinology has shaped her professional identity far beyond technical expertise. The specialty has instilled patience, humility, and a commitment to accompanying children and families through long-term care with empathy and continuity. She hopes to practise with curiosity and compassion, contributing meaningfully to the lives of those she serves. Fellowship has not only deepened her understanding of pediatric endocrinology but has also clarified the clinician she continues to grow into. What began as a period of training has become a way of thinking-moving forward not with all the answers, but with greater clarity in how to seek them.

In our regular feature on "Ped Endo Journal Scan," Sukanya Priyadarshini,<sup>[12]</sup> New Delhi, India, and Kriti Joshi, Brisbane, Australia, discuss six recent fascinating publications. The first is a study of non-autoimmune, insulin-deficient diabetes in children and young adults in Africa, describing evidence from the young-onset diabetes in sub-Saharan Africa cross-sectional study. The second review describes growth hormone withdrawal in mid-puberty, which shows no impact on near-adult height in adolescents with transient idiopathic growth hormone deficiency (GHD). The third study evaluates the diagnostic performance of morning serum cortisol for glucocorticoid weaning in children and adults. The fourth is a prospective, open-label study of calcitriol alone for the treatment of children and adults with X-linked hypophosphatemia. The fifth study reviews the benefits of longitudinal characterization and sonographic staging of testicular adrenal rest tumors. The sixth publication describes comprehensive molecular studies in Japanese patients with congenital hypogonadotropic hypogonadism. The editors hope that the compilation of these new research publications will provide insights into 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 will be grateful for your comments and suggestions, and we welcome contributions to the forthcoming issues of our journal.

Happy reading!

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