

Editorial

Editor's Page

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We welcome you to another interesting issue of our journal, which includes a compilation of original articles, case reports, reviews, and journal updates. We hope that these articles are academically exciting to the readers, especially the postgraduate students and fellows in training.

The editorial board would like to place on record the unstinted support that they have received from the outgoing Executive Council of ISPAE 2023–2024 under the leadership of Dr. Ahila Ayyavoo, President; Dr. Rakesh Kumar, Secretary-cum-Treasurer and Dr. Sirisha Kusuma, Joint Secretary. We look forward to another exciting and productive tenure under the new Executive Council for 2025–2026 led by Dr. Anurag Bajpai, President; Dr. Ravindra Kumar, Secretary; Dr. Saurabh Uppal, Joint Secretary; and Dr. Jaivinder Yadav, Treasurer.

In an original article in this issue, *Pinar Algedik et al.*^[1] from Istanbul, Turkey describe the psychological effects of precocious puberty (PP) in a cohort of Turkish girls presenting with central precocious puberty (CPP) and premature thelarche (PT). This study provides insight into the relationship between early pubertal onset and cognitive and emotional disturbances in the affected children and their mothers. The authors emphasize that evaluation of the psychological well-being is essential before initiating puberty-suppressing treatment with gonadotropin-releasing hormone analogs (GnRHa). While the benefits of GnRHa therapy in CPP on pubertal development are well recognized, there is a relative paucity of evidence regarding its effects on psychological status. A comprehensive clinical assessment and professional guidance from an endocrinologist can contribute to alleviating negative emotions and anxiety experienced by both the child and parents with CPP and PT over time.

Sameera Auckburally and Indraneel Banerjee from Manchester, UK in an accompanying editorial commentary^[2] emphasize the necessity for open communication with children and their families in the context of PP and the requirement for formal psychological counseling and review in addition to medical therapy. Negative perceptions regarding body image, lower self-esteem, and higher levels of depression were observed in girls with PP from Korea, China, and other regions. A limitation of the study by Algedik *et al.*,^[1] is its small sample size and lack of longitudinal follow-up. Clinicians can provide truly comprehensive care for children with PP only through recognition of both psychological and physical attributes of PP.

Ambica Tandon and Vijayalakshmi Bhatia from Lucknow, India note that studies^[3] of the psychological burden associated with PP and its possible alleviation with GnRHa therapy remain inconclusive. There are limited data on the psychological effects of early puberty in children and no formal data on PP from India. Parents frequently express concerns about the possibility of abuse, ability of girls to cope with menstruation, issues regarding future fertility, and potentially compromised final adult height. Pediatric endocrinologists should be cognizant of the social and

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psychological associations and implications of both early normal puberty and PP.

Alankrita Goswami and Sangita Yadav, New Delhi, India in a narrative review^[4] discuss the functional significance of maternal and neonatal vitamin D deficiency. Neonatal vitamin D insufficiency has been linked with preterm birth, seizures, neonatal respiratory distress syndrome, sepsis, acute gastroenterocolitis, and a higher risk of hospital admissions. The potential underlying mechanisms include the effect of vitamin D receptor polymorphism, defective immune effector cells, placental inflammation, and gut dysbiosis. Nearly half of all neonatal hypocalcemic seizures are attributable to vitamin D deficiency. The authors also examine the benefit of vitamin D supplementation during pregnancy on neonatal growth. Routine maternal vitamin D supplementation may be beneficial for optimal health of the newborns.

In “Case Series,” *Khubaib Ahmed et al. from Leeds, UK* describe four cases of 17-beta-hydroxysteroid dehydrogenase type 3 (17-β-HSD3) deficiency, a rare cause of disorders of sex development leading to an external female phenotype or varying degrees of undervirilization in 46,XY patients, presenting at different ages with distinct complaints. The authors review the clinical, biochemical, and genetic characteristics of children diagnosed with 17-β-HSD3 deficiency in a single tertiary center.^[5]

We have a few interesting “Case Reports” too. Pituitary stalk interruption syndrome (PSIS) is a rare condition characterized by multiple pituitary hormone deficiencies and a triad of aplastic or hypoplastic anterior pituitary, ectopic posterior pituitary, and interrupted pituitary stalk on imaging. *Shamkiran et al. from Bengaluru, India* report a case of a boy who presented with hyponatremic seizures and was subsequently diagnosed with PSIS, with a heterozygous mutation in the roundabout guidance receptor 1 gene.^[6]

Ganesh Jevalikar et al. from New Delhi, India describe the case of a boy who presented with rickets and was suspected to have nutritional vitamin D deficiency rickets precipitated by the use of anticonvulsant medications. The presence of proteinuria and lack of normalization of serum alkaline phosphatase prompted further evaluation which confirmed Dent disease type 2 secondary to mutations in the *OCRL1* gene.^[7]

Md Ejaz Alam et al.^[8] from *Srinagar, India* report a novel *STAG3* homozygous missense variant, c.926T>C (p.Phe309Ser), in a girl presenting with delayed menarche, features of gonadal dysgenesis, and 46,XX karyotype. This variant, located in the regulator of chromosome condensation 1 domain of the *STAG3* protein, probably disrupts the function of cohesin complex in meiosis, leading to premature depletion of ovarian follicles and primary ovarian insufficiency.

In the section on “Images and Spotters,” *Anju Bala et al.*^[9] from *Chandigarh, India* describe characteristic clinical and

radiographic images of a child with pycnodysostosis, a genetic disorder where the prominent clinical features include short stature with abnormal bone density, characteristic craniofacial features, and brittle bones.

In the section “Fellow’s Corner,” *Pamali Mahasweta Nanda, New Delhi, India* scripts her reflections on her journey through training in pediatric endocrinology.^[10] Opportunities for admission to a formal training program in a pediatric endocrinology in India are relatively scarce and limited to a few institutions. We hope that her experience and advice will be useful to those considering a career in pediatric endocrinology.

In our regular feature on “Ped Endo Journal Scan,” *Kriti Joshi, Brisbane, Australia*, discusses five recent fascinating publications.^[11] The first is a study on the safety and efficacy of once-daily oral therapy with infigratinib, an orally bioavailable fibroblast growth factor receptor 3 selective tyrosine kinase inhibitor in children with achondroplasia between the ages of 3 and 11 years – a new oral therapeutic option. The second review highlights the treatment regimens and glycemic outcomes in more than 100,000 children with type 1 diabetes during the period 2013–2022 in a longitudinal analysis of data from pediatric diabetes registries. The study demonstrated an improvement in mean hemoglobin A1c initiative during the 10-year study period. The third study summarizes the results of the first clinical trial of setmelanotide in patients aged 2–5 years with rare MC4R pathway-associated obesity (VENTURE) in a 1-year, open-label, multicenter, phase 3 trial. The fourth commentary abridges the impact of lomitapide for the treatment of pediatric patients with homozygous familial hypercholesterolemia (APH-19) from the efficacy phase of an open-label, multicenter, and phase 3 study. There was a significant overall reduction in LDL cholesterol after 24 weeks of treatment and an acceptable safety profile. The fifth report is on association of loss-of-function growth hormone (GH) secretagogue receptor (GHSR) variants with short stature and low insulin-like growth factor-1 (IGF-1). The observations of proportionate short stature, low IGF-1, and good response to recombinant human GH are in line with the reported function of GHSR and fit with the hypothesis of GH neurosecretory dysfunction in patients carrying pathogenic variants. The editors hope that these new research publications 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!

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