EDITORIAL

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Congenital Adrenal Hyperplasia and Adrenal Insufficiency in Children: An Evidence-based Review with Good Practice Points by Adrenal Working Group of The Turkish Society for Pediatric Endocrinology and Diabetes

Zeynep Şıklar

Ankara University Faculty of Medicine, Department of Pediatric Endocrinology, Ankara, Turkey

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Adrenal gland diseases in pediatric cases constitute a heterogeneous group of diseases. Within this group, congenital adrenal hyperplasia (CAH) and adrenal insufficiencies are important endocrine problems that require accurate diagnosis and urgent treatment. Recent developments, the development of genetic diagnostic methods, the widespread use of screening programs and innovations in the field of treatment have caused more attention in this diagnostic group.

Fifteen years ago, various working groups according to endocrine disease groups were established within the Turkish Society for Pediatric Endocrinology and Diabetes. 'Adrenal Working Group' is one of these groups with the aim of improving the care of children with pediatric adrenal diseases. This working group plans and continues educational activities such as preparing diagnosis and treatment recommendations for pediatric adrenal diseases, preparing materials (videos, brochures) to inform pediatricians and pediatric endocrinologists, and organizing seminars and conferences.

Classical CAH is the most common form of primary adrenal insufficiency in childhood and is a potentially life-threatening condition. In our country, a pilot study found that the incidence of classical 21-hydroxylase deficiency in the screened population was 1:7,787; subsequently, the initiation of the newborn CAH screening program was a significant diagnostic step (1).

Moreover, since most diseases of the adrenal gland are hereditary, their incidence is higher in societies where consanguineous marriages are common than in other societies. Studies reported from our country draw attention to the importance of adrenal problems (2,3,4,5,6). In addition to common diseases of the adrenal gland, sharing experiences with rare adrenal gland problems will also be useful for the management of these diseases.

One of the rare causes of CAH is 17-hydroxylase deficiency. A publication from our country that included the largest pediatric endocrine case series, analyzed data from a total of 97 cases from 78 families. In addition to important clinical and genetic data specific to 17-hydroxylase deficiency, data

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Address for Correspondence: Zeynep Şıklar MD, Prof, Ankara University Faculty of Medicine, Department of Pediatric Endocrinology, Ankara, Turkey
E-mail: zeynepsklr@gmail.com **ORCID:** orcid.org/0000-0003-0921-2694

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from a significant number of patients regarding the final height of the cases were presented (7).

New medications are being introduced for adrenal gland diseases, especially for CAH. Children with classic CAH require treatment with glucocorticoids, usually at supraphysiologic doses, to address cortisol insufficiency and reduce excess adrenal androgens. However, such treatment confers a predisposition to glucocorticoid-related complications. For this reason, there are new treatment interventions such as corticotropin-releasing factor type 1 receptor antagonist, modified release hydrocortisone etc. (8,9).

Despite all the developments and advances, the diagnosis and treatment of adrenal insufficiency remains a challenge for both patients and healtcare providers. Some problems are occasionally encountered in those cases. It is known that, patients with adrenal insufficiency have increased morbidity, mortality and impaired quality of life (1). Understanding the issues that may arise during long-term follow-up is another important aspect of managing these patients.

It is very valuable for physicians working in the field of pediatric endocrinology, as well as pediatricians who first encounter patients, to have access to adequate information about CAH and adrenal insufficiency, especially in managing patients during emergencies.

It has been acknowledged by the 'Adrenal Working Group' that there is a need to prevent these difficulties, raise awareness about CAH and adrenal insufficiency, and create an easily accessible and holistic resource in diagnosis and treatment. This evidence-based review with good practice points developed by 'Adrenal Working Group of 'The Turkish Society for Pediatric Endocrinology and Diabetes' to provide good practice points, with focus on recommendations for daily management of adrenal diseases including CAH and adrenal insufficiencies.

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