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SYMPTOMATOLOGY OF CONGENITAL ADRENAL HYPERPLASIA DUE TO 21-HYDROXYLASE ENZYME DEFICIENCY

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Abstract: Introduction: Deficiency of the enzyme 21-hydroxylase (21-OH) is responsible for approximately 90% of cases of Congenital Adrenal Hyperplasia (CAH) due to its impact on the cortisol biosynthesis pathway. It is presented by the classic form and non-classical form, also called late expression. Disease severity is related to the degree of 21-OH enzyme activity that is suppressed by mutation in the CYP21A2 gene. The classic form is characterized by clinical manifestations generated by the excess of androgens produced by the deviation of the cortisol metabolic pathway. In late-expression CAH, the enzymatic blockade is partial and massive cortisol deficiency does not occur. **Objectives.** Elucidate CAH caused by 21-OH deficiency and its possible classic and non-classic symptoms, highlighting the complications generated by not performing early treatment. **Methodology.** The study in question is a literature review without meta-analysis based on data from Pubmed, Scielo and Google Scholar, using 2 articles in Portuguese and 1 in Spanish. The following descriptors were used: Congenital adrenal hyperplasia; 21-hydroxylase enzyme; Cortisol. **Results and discussion.** CAH causes an overproduction of corticotropin-releasing hormone in the hypothalamus and adrenocorticotropic hormone by the pituitary gland, stimulating the adrenal corticosteroid pathway and causing hyperandrogenism. Failure to treat patients who have CAH due to 21-OH deficiency has the following harms: In the classic, untreated form in males, it causes progressive postnatal virilization with obvious signs and symptoms of precocious puberty, such as acne, penile enlargement without enlargement testicular, skeletal maturation leading to short stature and decreased male fertility may occur. In females, prenatal virilization of the external genitalia is reported, which may progress to progressive masculinization of the genitalia in the postnatal period, menstrual irregularities, advancing bone age with short stature, and may be related to symptoms similar to polycystic ovary syndrome, due to high levels of androgens. The non-classical form allows partial activity of the cortisol metabolic pathway and the signs of hyperandrogenism are milder than in the classic untreated forms in both sexes. In addition, its manifestations are later and occur with the early onset of pubic hair, advancing bone age and acne, which can be mitigated over time or even go unnoticed. In adult men, the non-classical form can develop early baldness, low fertility, oligozoospermia and small testicles in relation to the size of the phallus. **Conclusion.** CAH is the main pathology caused by 21-OH enzyme deficiency and causes severe clinical manifestations in both affected sexes, mainly in the classic form. Therefore, further studies on the subject are needed to better understand the symptoms and enable health professionals to have a favorable conduct for the prognosis of the disease and to control the aforementioned complications.

Keywords: Congenital Adrenal Hyperplasia; 21-Hydroxylase; Adrenal; Cortisol.

REFERENCES

DELAI, A. Avaliação do perfil cardiometabólico na forma não clássica da hiperplasia adrenal congênita por deficiência da 21-hidroxilase. 2021. Tese (Doutorado em Clínica Médica) - Faculdade de Medicina de Ribeirão Preto, Universidade de São Paulo, Ribeirão Preto, 2022.

PRADO, M.J. Hiperplasia adrenal congênita: predição e elucidação do efeito de mutações no gene CYP21A2 humano na estrutura e na atividade da enzima 21-hidroxilase. 2022.

Tese (Doutorado em Biologia Celular e Molecular) - Universidade Federal do Rio Grande do Sul, Porto Alegre, 2022.

SÁNCHEZ, C.M.J. et al. Tendencias actuales de la hiperplasia adrenal congénita, una revisión de la literatura. Journal of American Health, v. 5, n. 2, 2022.