Papulocistic lesions of the face: the tip of the iceberg

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Abstract

Acne is a manifestation of hormonal overstimulation of the pilosebaceous units of genetically susceptible individuals and may manifest in the form of comedonic, papulopustular or nodular lesions. It can present as an isolated disease or in the context of clinical syndromes; among these congenital adrenal hyperplasia (CAH),a group of autosomal recessive disorders resulting from deficiency of one of the five enzymes required for synthesis of cortisol in the adrenal cortex.

The most common form of the disease is classic 21-hydroxylase deficiency, which is characterized by decreased synthesis of glucocorticoids and often mineralocorticoids, adrenal hyperandrogenism and impaired development and function of the adrenal medulla. The impaired glucocorticoid feedback inhibition at the hypothalamic and anterior pituitary levels leads to increased secretion of corticotropin-releasing hormone (CRH) and adrenocorticotropic hormone (ACTH) respectively, adrenal hyperplasia, and increased production of adrenal androgens and steroid precursors prior to the enzymatic defect . The clinical spectrum of 21-hydroxylase deficiency is quite broad, ranging from the most severe to mild forms, depending on the degree of 21-hydroxylase. Here we report the case of a young woman affected by acne refractory to treatment as the only sign of CAH.

Keywords: *Congenital adrenal hyperplasia, 21-hydroxylase deficiency, CRH, PCOS, hormonal disorders, Acne*

Case presentation

A 20-year-old girl presented to our acne outpatient service because of papular and post inflammatory lesions of her face refractory to both topic and systemic treatment. She referred the onset of papulopustular lesion of her face associated with dysmenorrhea and mild signs of hyperandrogenism since the last two years (mild hypertricosis). The medical history was positive for polycystic ovaric syndrome (PCOS). In the family history her mother suffered from acne and diabetes, while her father died of stroke. Given the family history for cardiovascular risk factors, treatment with estrogen and progestin was previously delayed. She referred us she had been prescribed treatment with inositol instead, with partial control of dysmenorrhea, without any resolution of acneic lesions.

Therefore she was set to antibiotic therapy for 12 weeks unsucesfully. Eventually, considering the risk-benefit profile, she was prescribed estroprogestinic treatment, without benefit again. So she was referred to another dermatologist, prescribing her oral isotretinoin at dosage of 0.5mg/kg daily for 16 weeks.

This treatment improved her clinical condition, but she underwent relapse three months after the suspension of it. At this point she came to our attention, and we decided to revaluate the case, given the previous terapeutic failure. The patient underwent terapeutic wash-out for sistemic treatment and after one month was addressed to an endocrinologist for a complete study of her hormonal background.

The laboratory tests resulted in the normal range, except for a slight alteration of cortisol. This suggested the performing of further tests, such as the stimulus test with ACTH for 17alpha-hydroxyprogesterone, which turned positive, suggesting the diagnosis of non classical adrenogenital syndrome. Given this diagnosis she was set on sistemic steroid treatment by her endocrinologist, in meantime she continued topical therapy for acne with benefit.

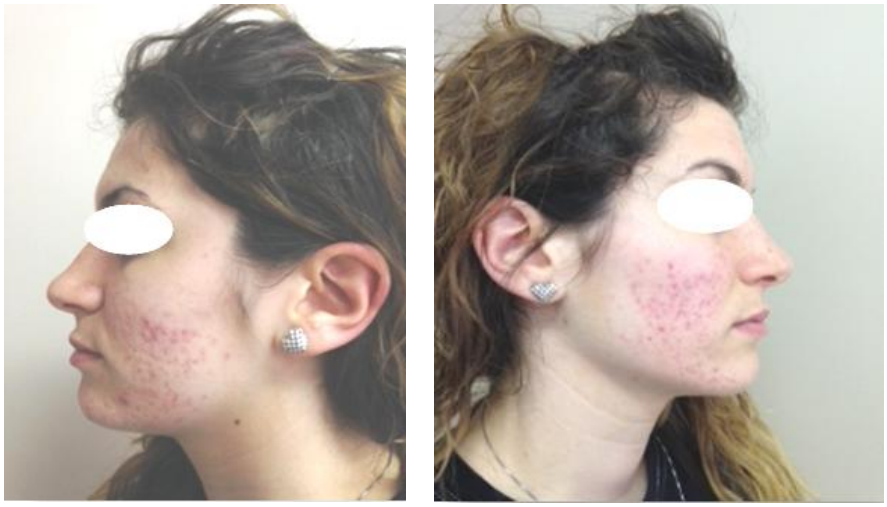

Figure 1. Before diagnosis and treatment.

Conclusion

Recent data reported that acne is the most common skin disorder seen in ambulatory dermatology practices regardless of gender, skin color or ethnicity [1]. Female patients are more likely to develop anxiety and depression due to their condition, and acne improvement positively influences quality of life. The patient's sex should not radically alter diagnostic or therapeutic efforts, although gender differences could be necessary to set up clinical management, monitoring also the psychological aspect [2].

Figure 2. Afer diagnosis and treatment.

The findings of this report, according to literature emphasize that acne is not merely an inflammatory disease of skin, but it can be a possible indicator of clinical syndromes or the expression of genetic disorders, such as CAH [3].

This is a group of genetic disorders in cortisol biosynthesis, most commonly steroid 21-hydroxylase deficiency (21OHD) and acne can be a clinical manifestation of these defects [4]. For this reason complex clinical forms of acne and/or refractory ones to treatment should prompt further laboratory and instrumental investigation. This case once again confirms the need for interdisciplinary integration in the study of very complexive acne cases.

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