Case report

Aperts Syndrome with Intractable Acne Vulgaris

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INTRODUCTION

Apert syndrome is an autosomal dominant disorder first described in 1906. Its incidence ranged from 1/160,000 to 200,000 live births [1], and linked with high parental age. It is caused by mutation in fibroblast growth factor (FGFR-2), and occurs as a result of androgen hyper response affecting the epiphysis and sebaceous glands [2]. This syndrome is characterized by craniosynostosis (premature fusion of cranial sutures), facial malformation, midface, hypertelorism, and laterally down sloping slanting eyes. Additionally, patients may display a small nose, low-set ears, symmetrical limb syndactyl (cutaneous and bony fusion of the digits) and a variety of abnormalities including skin, brain and visceral organs [3,4].

Acneiform lesions in patient with apert syndrome were first described by Solomon in 1971[5]. Oily skin is noted at adolescence, with subsequent appearance of acne papules, the distribution of lesions is more diffuse, often involving the forearms, buttocks, and thighs. The etiology is hypothesizing either to the end-organ androgen metabolism defects which may lead to sebaceous gland abnormalities. Reports have revealed that FGFR-2 might play a role in regulating androgen sensitivity of the Folliculo-sebaceous unit [6].

CASE REPORT

A 14-year old male with clinical features of a pert syndrome presented with sudden history of multiple comedones, papules, pustules on the face, arms and upper trunk which began at age of 10 years old and persist over 4 years. The child born after 36 weeks of
gestation by caesarean section. Presented with multiple congenital anomalies, systemic examination revealed no abnormalities early surgery of skull done at age of 3 months to detach skull plate from one another and to relieve cranial pressure. Disclosed blindness at age of 10 year, his language and social development was compatible with age. Both parents were normal, has three normal siblings. No family history of similar complaint or any other congenital abnormalities.

A standard management with intermittent courses of local antibiotics, benzoyl peroxide and tretinoin and systemically with antibiotics (Tetracycline, Erythromycin) did not result in any improvement. Lastly, the case was treated with 20mg isotretinoin daily for seven days every month for a total period of 4 months with good result.

**Figure 1.** Abnormal head contour, hypertelorism, eye bulging and down sliding of lateral palpebral fissures malocclusion teeth, fused fingers and fused toes.

**Figure 2.** Scattered comedons, papules, pustules on face, arms and trunk.

**Figure 3.** Patient after isotretinoin treatment.

**DISCUSSION**

The importance of this report is to present a significant clear association between a pert’s syndrome and acne vulgaris with resistance to usual acne treatment. Our finding supports previous reports suggests a role of isotretinoin as effectively treats all forms of acne vulgaris [7].

**Declaration of patient consent**

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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**Conflicts of interest**

There are no conflicts of interest.

**REFERENCES**


