

*Case report*

# Aperts Syndrome with Intractable Acne Vulgaris

Fauzia Musbah<sup>1,2</sup> <sup>1</sup> Department of Dermatology, Faculty of Medicine, University of Tripoli, Tripoli, Libya.<sup>2</sup> Department of dermatology, Tripoli Central Hospital, Tripoli, Libya**ARTICLE INFO**DOI: [10.5281/zenodo.3938709](https://doi.org/10.5281/zenodo.3938709)\* **Fauzia Musbah:** Department of Dermatology, Tripoli Central Hospital, Tripoli, Libya. **Mobile phone:** (+218) 925007361. **Email:** [fauziamusbah@yahoo.com](mailto:fauziamusbah@yahoo.com)**Received:** 07-06-2020**Accepted:** 11-06-2020**Keywords:** Apert syndrome, Craniosynostosis, Acne vulgaris.

This work is licensed under the Creative Commons Attribution International License (CC BY 4.0).

**ABSTRACT**

A pert syndrome as rare congenital disorders characterized by craniofacial and limb abnormalities. A 14 years old boy presented with a severe acne vulgaris for 4 years. In this case report, we discuss this rare condition and its treatment option.

**Cite this article:** Fauzia Musbah. Aperts Syndrome with Intractable Acne Vulgaris. *Alq J Med App Sci.* 2020;3(2)7-9.

**INTRODUCTION**

Apert syndrome is an autosomal dominant disorder first described in 1906. Its incidence ranged from 1/160,000 to 200,000 live births<sup>[1]</sup>, 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<sup>[2]</sup>. 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<sup>[3,4]</sup>.

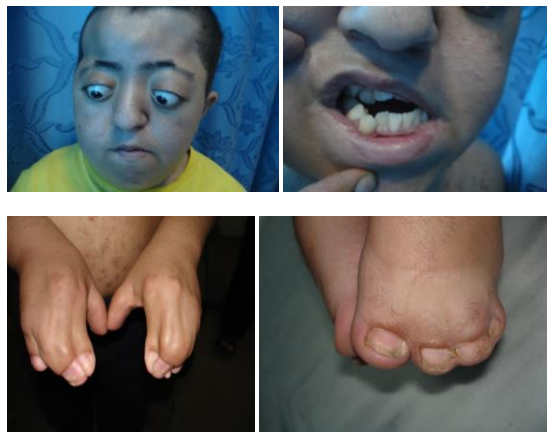
Acneiform lesions in patient with *apert* syndrome were first described by Solomon in 1971<sup>[5]</sup>. Oily skin is noted at adolescence, with subsequent appearance of acne papules, the distribution of lesions is more diffuse, often involving the forearms, buttocks, and thighs. The etiology is hypothesizing 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<sup>[6]</sup>.

**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.



**Figure1.** Abnormal head contour, hypertelorism, eye bulging and down sliding of lateral palpebral fissures malocclusion teeth, fused fingers and fused toes.



**Figure2.** Scattered comedons, papules, pustules on face, arms and trunk.



**Figure3.** 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.

### *Financial support and sponsorship*

Nil.

### *Conflicts of interest*

There are no conflicts of interest.

## REFERENCES

- [1] Harper JI. Genetics and genodermatosis. In: Champion RH, JL, Breathnach SM, editors. Rook/wilkinson/Ebling Textbook of dermatology. 6th ed. Oxford: Blackwell Science. 1998:425-6.9.
- [2] Wilkie AO, Slaney SF, Oldridge M, Poole MD, Ashworth GJ, Hockley AD, et al. Apert syndrome results from localized mutations of FGR2 and is allelic with Crouzon syndrome. Nat Genet 1995;9:165-172.

- [3] Blank CE. Apert's syndrome (a type of acrocephalosyndactyly): observations on a British series of thirty-nine cases. *Ann Hum Genet* 1960; 24:151-163.
- [4] Robin NH, Falk MJ, Haldeman-Englert CR. FGFR-Related craniosynostosis syndromes. In: Adam M.P., Ardinger HH, Pagon RA, editors. *Gene Reviews* [Internet] University of Washington, Seattle; Seattle (WA): 1998.
- [5] Solomon LM, Fretzin D, Pruzansky S. Pilosebaceous abnormalities in Apert's syndrome. *Arch Dermatol* 1970;102:381-285.
- [6] Henderson CA, Knaggs H, Clark A, Highet AS, Cunliffe WJ. Apert's syndrome and androgen receptor staining of the basal cells of sebaceous glands. *Br J Dermatol* 1995; 132:139-143.
- [7] Benjamin LT, Trowers AB, Schachner LA. Successful acne management in Apert's syndrome twins. *Pediatr Dermatol* 2005; 22:561-565.