Indian Journal of Medical Case Reports ISSN: 2319–3832
Online, International Journal, Available at http://www.cibtech.org/jcr.htm
2022 Vol.11, pp. 27-28/Navyatha et al.
Case Report (Open Access)

A RARE AND SPORADIC CASE OF ADAMS - OLIVER SYNDROME: A CASE REPORT

***Chamarthi Navyatha, A. Sumalatha and A. Vijayamohan Rao**
Department of Dermatology, Venereology, Leprology, Narayana Medical College, Nellore
**Author for Correspondence: navyachamarthi22@gmail.com*

ABSTRACT
Adams - oliver syndrome is a rare inherited condition present at birth that involves changes to limbs and scalp. Symptoms may include areas of missing skin on scalp, limb abnormalities, heart defects, neurological concerns and issues with the eyes.

Keywords: *Adams-Oliver Syndrome, Congenital Disorder*

INTRODUCTION
Adams oliver syndrome is a variably expressed congenital disorder which is considered to be very rare. Incidence is about 1 in 225,000 live births. It is characterized by the presence of Aplasia cutis congenita of the scalp and transverse limb defects. It is classified as Type 2 Aplasia cutis congenita under Frieden's classification. It is usually inherited as autosomal dominant or autosomal recesssive form with occasional sporadic cases (Mendiratta *et al.*, 2017).

CASE
A 14 year old male presented with cicatricial alopecia of the scalp since 2-3 months of age. History of presence of large raw areas over the scalp along with defects in the fingers of both hands since birth. The raw areas later healed with hairless scars. He is the only child born of a non-consanguinous marriage delivered at full term through LSCS. No history of fluid filled lesions prior to the onset of raw areas. No history of similar complaints in the family. Unilateral left undescended testes was detected on ultrasonography and underwent left laparoscopic orchidopexy. On examination, four smooth shiny patches of cicatricial alopecia noted over the vertex, brachydactyly of both hands, low set ears and high arched palate were present. Toes were normal. No other lesions were noted elsewhere on the body.

Figure 1 & 2: Aplasia cutis

Figure 3: Low set ears

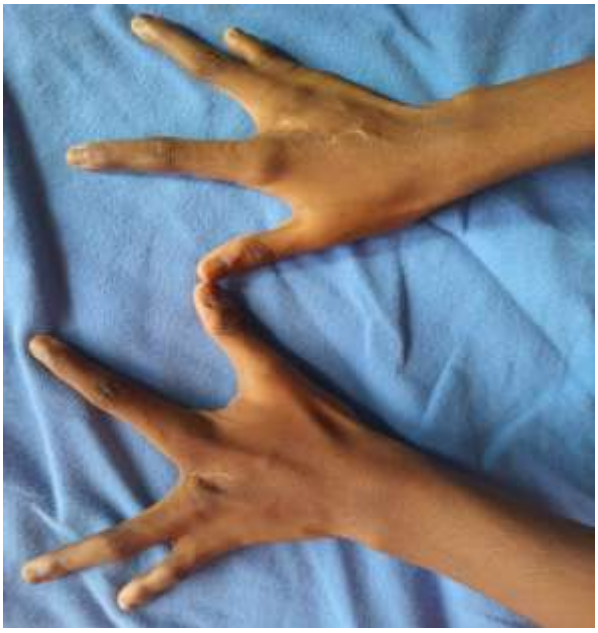

Figure 4: Brachydcatyly

DISCUSSION

Adams – oliver syndrome is a rare condition characterized by various malformations of the limbs and abnormal skin development, particularly on the scalp (Mendiratta *et al.*, 2017). The aetiology is unkown. However, family history is a risk factor; a parent who is a carrier has 50% chance of passing this syndrome to offspring (Saeidi and Ehsanipoor, 2017). The risk appears to be same for males and females. Adams – oliver syndrome is caused by genetic mutations in at least 6 genes: ARPGAP31, DLL4, DOCK6, EOGT, NOTCH1, or RBPJ. Each of these genes plays a pivotal role in embryonic development. Inheritance pattern can be autosomal dominant or less commonly, autosomal recessive. Diagnosis is based on clinical signs and symptoms, patient / family history and imaging.

CONCLUSION

We are reporting this case because it has occurred sporadically as our patients family had no congenital scalp or limb defects and as it is a rare case with limited cutaneous expression highlighting the varied phenotypic expression of Adams-oliver syndrome.

REFERENCES

Mendiratta V, Marak A, Chander R and Yadav A (2017). Adams–Oliver syndrome: Report of a sporadic case with limited cutaneous expression. *Indian Journal of Paediatric Dermatology*, **18**, 46-92.

Saeidi M and Ehsanipoor F (2017). A Case of Adams-Oliver Syndrome. *Advanced Biomed Research,* **6**, 167. doi:10.4103/2277-9175.221861