ECRONICON OPEN ACCESS

EC OPHTHALMOLOGY
Case Report

Case Report: Phocomelia with Coloboma Dextrocardia

Nilanjan Chowdhury[1], Sanjoy Chowdhury[2]*, Priti Sinha[3] and Sneha Kumari[3]

[1]*Department of Ophthalmology (PGT), Kolkata Medical College, Kolkata, India*
[2]*tt7E3 Joint Director (Medical and Health Services), Jharkhand, India*
[3]*DNB Residents, Bokaro General Hospital, Jharkhand, India*

***Corresponding Author:** Sanjoy Chowdhury, tt7E3 Joint Director (Medical and Health Services), Jharkhand, India.

Received: November 26, 2021; **Published:** March 31, 2022

Abstract

Phocomelia is caused by symmetrical limb reduction in utero. Syndromes associated with include Robert's syndrome, Thrombocytopenia with absent radius syndrome, Waardenburg syndrome and Holt-oram syndrome. Also, in utero exposure of thalidomide, alcohol and cocaine can cause similar musculoskeletal deformities. We report a case of phocomelia with total complete coloboma in both eyes along with dextrocardia in a 35 year old male. Due to lack of any significant history of drug abuse in mother of our patient and lack of associated signs of other syndrome, our case appear to be a result of a sporadic non-hereditary limb deficiency with typical total coloboma (typical coloboma of iris, choroid and retina) Presentation in middle age makes this case more unique.

Keywords: *Phocomelia; Robert's Syndrome; Thrombocytopenia; Waardenburg Syndrome; Holt-Oram Syndrome*

Introduction

Phocomelia is an extremely rare condition of malformation of arms and legs. Derived from Greek words phoka and melos meaning seal and limb respectively. Besides genetic aetiology, use of thalidomide has played an important role in its causation. Presenting features include underdeveloped limbs and absent pelvic bones. According to NORD (National Organization of Rare Disease) individuals with phocomelia syndrome generally show symptoms of growth retardation in-utero.

Case Presentation

We report a case of an adult male with symmetrical upper limb reduction associated with typical total coloboma and dextrocardia without any other typical features of phocomelia syndrome. A 35 year old male came for regular check-up of his glasses without any other ocular complaints. General physical examination was un remarkable but patient refused consent for further imaging and genetic testing. However he gave consent for photograph and publication.

Examination

He has underdeveloped arms, forearms, fingers and absent thumb in both upper limb. Also, he had total typical coloboma in both the eyes involving iris, choroid and retina. Vision was 6/6 in both eyes. Other systemic examination was within normal limits except that he has dextrocardia. His mother reported no use of any drug during gestation and that delivery was normal and baby was not low weight.

Figure 1: *Short arm, forearm, and fingers and absent thumb in both upper limb.*

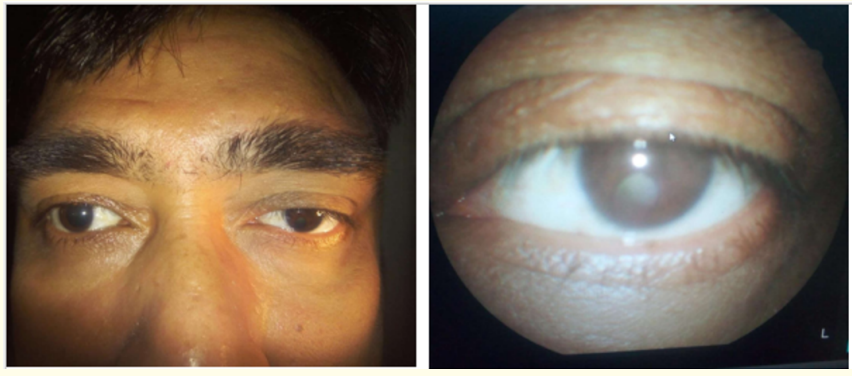

Figure 2: *Total typical coloboma in both eyes.*

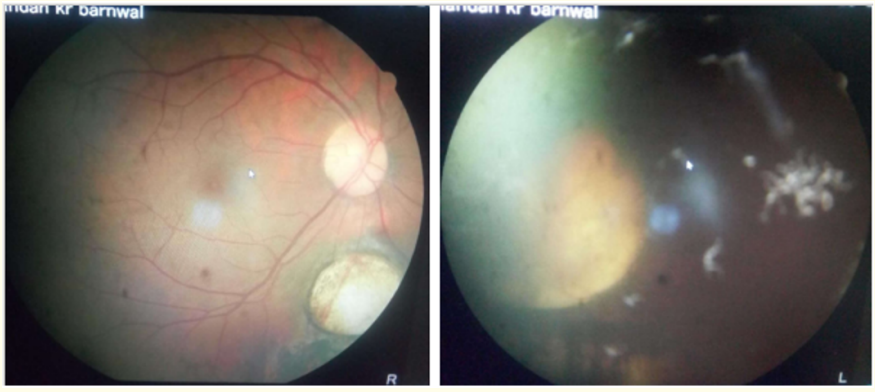

Figure 3: *Fundus photograph showing total coloboma in both eyes.*

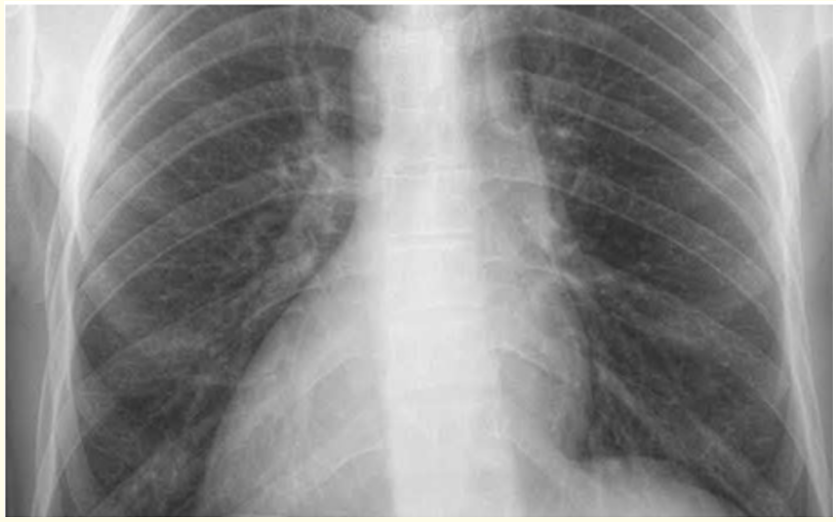

Figure 4: *X- ray showing dextrocardia.*

Discussion and Conclusion

Tetra phocomelia is a severe combination of limb defects in which total or partial agenesis of upper and lower limbs is seen, leading to proximity of limbs to the trunk. Such babies were termed thalidomide babies, a consequence of dysmorphogenesis of limb buds due to the use of thalidomide during 1960s. Now, X-ray irradiation, genetic and sporadic anomalies has also been attributed as its cause. Thalidomide was withdrawn in 1961.

Phocomelia syndrome is characterised by underdeveloped limbs and absent pelvic bones. Usually, the upper limbs are not fully formed and sections of hands and arms may be missing. Short arm bones and missing thumbs often occur, as evident in our case.

In phocomelia, the cells cease to develop or die, thus preventing proper development of limbs, eyes, palate, brain or other structures [1-3]. Involvement of eye is manifested as bilateral total typical coloboma in our case.

Differential diagnosis

Robert's syndrome: Extremely rare disorder, caused by malformation of bones in face, skull, arms and legs [4]. Thrombocytopenia-absent radius syndrome: caused by low platelet count, an absent radius, a hypoplastic thumb, and cardiac anomaly [5]. Holt-oram syndrome: autosomal dominant; caused by abnormal limb development affecting mostly forearm and the carpal bones. Three fourth patients have atrial septal defect or ventricular septal defect [6].

Bibliography

1. National Organization for Rare Disorders. Phocomelia syndrome.
2. Shekokar SP., *et al*. "Phocomelia: an extremely rare congenital disorder involving the limbs (dysmelia)". *International Journal of Infertility and Fetal Medicine* 2 (2011): 118-120.
3. Chakre GS., *et al*. "Phocomelia syndrome a case report". *Journal of Krishna Institute of Medical Sciences University* 1.2 (2012): 150-151.
4. Gordillo M., *et al*. "Roberts Syndrome". In: Pagon RA, Adam MP, Ardinger HI-1, Wallace SE, Amemiya A, Bean LJI-1, *et al*. editors. Gene Reviews.
5. Toriello HV. "Thrombocytopenia Absent Radius Syndrome". In: Pagon RA, Adam MP, Ardinger 1111, Wallace SE, Amemiya A, Bean UK., *et al*. editors. Gene Reviews.
6. Avina-Fierro JA and Colonnelli- Barba G. "Holt-Otani syndrome associated with facial anomalies. A case report". *Revista Médica del Instituto Mexicano del Seguro Social* 48.6 (2010): 657-659.

Volume 13 Issue 4 April 2022