ECRONICON OPEN ACCESS

EC OPHTHALMOLOGY
Case Report

Microphthalmia with Linear Skin Defect: A Clinical Diagnosis

Ritesh Kumar Shah[1]* and Raghunandan Byanju[2]

[1]*Bharatpur Eye Hospital, Bharatpur, Chitwan, Nepal*

[2]*Professor, Chief Medical Director and Head of Department, Department of Retina, Bharatpur Eye Hospital, Bharatpur, Chitwan, Nepal*

***Corresponding Author**: Ritesh Kumar Shah, Bharatpur Eye Hospital, Bharatpur, Chitwan, Nepal.

Received: October 30, 2018; **Published:** March 22, 2019

Abstract

Background: Microphthalmia with linear skin defect (MLS) syndrome is a rare X linked dominant disorder characterized by microphthalmos or anophthalmos and linear skin defect typically involving face and neck. Clinical findings unique to this disorder are usually sufficient to establish a diagnosis in circumstances where genetic analysis cannot be performed.

Case Presentation: We describe a 10-month-old female infant with unilateral anophthalmos and linear, atrophic skin lesion affecting face. Further examination revealed left orbital cyst, developmental delay and external ear anomaly. Genetic analysis could not be done as the mother refused further investigation in the child. The child was diagnosed with Microphthalmia with linear skin defect syndrome based on the presence of characteristic ocular and skin findings.

Conclusion: MLS syndrome can be diagnosed clinically based on the presence of characteristic ocular and skin finding and does not necessarily require a molecular analysis.

Keywords: *Anophthalmos; Linear Skin Defect; X Linked Disorder; Clinical Diagnosis*

Background

Microphthalmia with linear skin defect (MLS), also known as MIDAS (Microphthalmia, Dermal aplasia and Sclerocornea), is X-linked dominant neurocutaneous disorder with male lethality in utero. It is typically characterised by unilateral or bilateral microphthalmia and/or anophthalmia and linear atrophic skin streaks affecting face and neck.

MLS is a very rare disorder and only a handful of cases have been reported so far. We report an infant girl with typical features of MLS syndrome - unilateral anophthalmia and linear skin streaks involving the face.

Case Presentation

A ten-month-old female Mongolian child was brought by her mother for evaluation of a cystic mass in left orbit. According to the mother, the mass had been there since birth and had been gradually increasing in size. The child was born to non-consanguineous parents at home by normal vaginal delivery at full term. Her birth weight was normal. She was the fourth child and all her` brothers and sister were healthy.

Ocular examination revealed absent left eyeball with a cystic mass of size 2 x 2 cm in the infero-nasal aspect of the orbit with positive transillumination (Figure 1). Palpation of orbital rim revealed absent inferonasal orbital rim. She could follow light with her right eye although there was nystagmus. Cornea was clear and the posterior segment finding was unremarkable. Dermatological examination showed linear, depressed hypopigmented scar predominantly affecting left side of the face. The scar extended from the tragus of left ear

till nasal bridge passing over the left cheek (Figure 2). She also had a malformed left external ear (Figure 3). Systemic examination showed normal limbs, external genitalia, no cleft palate and had no teeth eruption. Motor and speech development was delayed as she could barely sit with support and only made sound while crying and laughing.

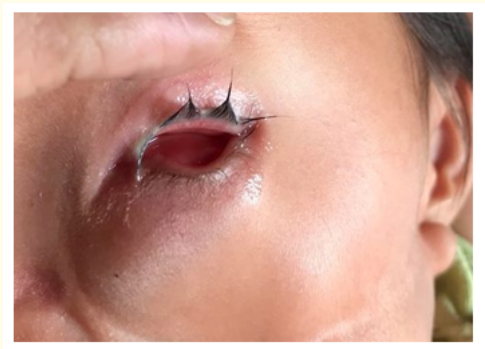

Figure 1: *Left primary anophthalmos with infero-nasal orbital cyst.*

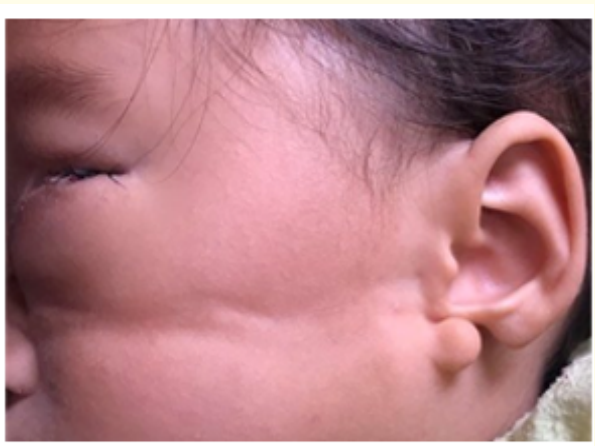

Figure 2: *Left malformed ear with linear depressed skin streak over the cheek.*

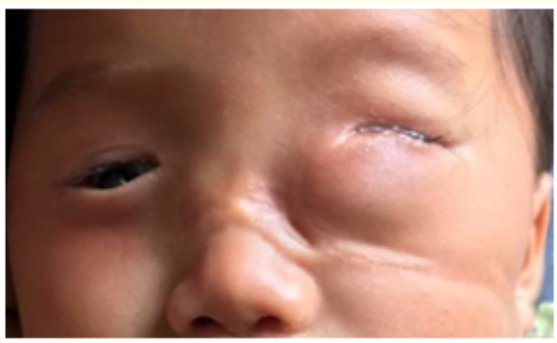

Figure 3: *Depressed linear skin streak involving left cheek and nasal bridge.*

A clinical diagnosis of Microphthalmia with linear skin defect was made based on presence of two major characteristic findings - anophthalmia and linear skin lesion involving face. This was further supported by features like orbital cyst, malformed ear and developmental delay. Cytogenetic analysis and systemic evaluation could not be done as the mother refused any further investigations and evaluation.

Discussion

Microphthalmia with linear skin defect was first described in 1988 [1]. Since then 62 cases with typical clinical findings had been reported till 2015 [2].

The clinical diagnosis of MLS syndrome can be established based on the presence of two major criteria (microphthalmia or anophthalmia and linear skin defect), however cases with established molecular diagnosis have reported only ocular or skin involvement [3]. Ocular findings other than microphthalmos or anophthalmos include sclerocornea, orbital cysts, microcornea, corneal leukoma, iridocorneal adhesion (Peters anomaly), congenital glaucoma, aniridia and cataracts [4,5]. Other occasional associations include central nervous system abnormalities, cardiac defects, developmental delay and genitourinary malformations. However, high inter- and intrafamilial phenotypic variabilities have been reported in these cases [4,6].

The most commonly observed chromosomal abnormality during cytogenetic analyses is monosomy for Xp22. Other abnormalities reported are XX karyotype with translocation of Y chromosome material on to X chromosome and XY male with a mosaic paracentric inversion of Xp [7,8]. In our case cytogenetic analysis was not done.

Other phenotypically similar conditions to MLS syndrome like Goltz syndrome, Oculocerebrocutaneous syndrome and Incontinentia pigmenti were considered but rejected based on history and clinical grounds.

Conclusion

Clinical diagnosis of MLS syndrome can be established based on typical ocular and dermatological features of this disorder which can be identified by any ophthalmologist, dermatologist or pediatrician. Where genetic analysis is not possible, it may not be done.

Declarations

Ethic Approval

An ethical approval was obtained from the hospital research committee.

Consent for Publication

"Written informed consent was obtained from the mother of the child for publication of this case report and any accompanying images. A copy of the written consent is available for review by the Editor-in-Chief of this journal."

Availability of Data and Material

Data sharing is not applicable to this article as no datasets were generated or analysed during the current study.

Competing Interests

The authors declare that they have no competing interests.

Funding

None.

Authors Contribution

- Dr Shah R was involved in clinical examination of the case and writing of the manuscript.
- Dr Byanju R was involved in reviewing the manuscript and providing expert opinion.

Acknowledgement

None.

Bibliography

1. Happle R., *et al*. "MIDAS syndrome (microphthalmia, dermal aplasia and sclerocornea): an X-linked phenotype distinct from Goltz syndrome". *American Journal of Medical Genetics* 47.5 (1993): 710-713.

2. Rahden A., *et al*. "Clinical spectrum of females with *HCCS* mutation: from no clinical signs to a neonatal lethal form of the microphthalmia with linear skin defects (MLS) syndrome". *Orphanet Journal of Rare Cases* 9 (2014): 53.

3. Morleo M and Franco B. "Dosage compensation of the mammalian X chromosome influences the phenotypic variability of X-linked dominant male-lethal disorders". *Journal of Medical Genetics* 45.7 (2008): 401-408.

4. Cape CJ., *et al*. "Phenotypic variation in ophthalmic manifestations of MIDAS syndrome (microphthalmia, dermal aplasia, and sclerocornea)". *Archives of Ophthalmology* 122.7 (2004): 1070-1074.

5. Allanson J and Richter S. "Linear skin defects and congenital microphthalmia: a new syndrome at Xp22.2". *Journal of Medical Genetics* 28.2 (1991): 143-144.

6. Kobayashi M., *et al*. "An XX male with microphthalmos and sclerocornea". *Journal of Pediatric Ophthalmology and Strabismus* 35.2 (1998): 122-124.

7. Morleo M., *et al*. "Microphthalmia with linear skin defects (MLS) syndrome: clinical, cytogenetic, and molecular characterization of 11 cases". *American Journal of Medical Genetics Part A* 137.2 (2005): 190-198.

8. Kutsche K., *et al*. "Microphthalmia with linear skin defects syndrome (MLS): a male with a mosaic paracentric inversion of Xp". *Cytogenetic and Genome Research* 99.1-4 (2002): 297-302.

Volume 10 Issue 4 April 2019