OPHTHALMO-ACROMELIC SYNDROME IN A PALESTINIAN INFANT: A CASE REPORT

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ABSTRACT

Ophthalmo-acromelic syndrome (OAS) is a rare congenital disorder that leads to malformations of the eyes and limbs. In this article, we have reported the case of a newborn girl with OAS. She was the fifth child in the family. The non-consanguineous parents were healthy. The newborn had bilateral true anophthalmia, macrodactyly and oligodactyly (two toes in both feet, three fingers in the left hand, and four in the right hand). In addition, she had lumbar spina bifida and hydrocephalus. This was not reported before in the same syndrome. The number of reported cases of this syndrome around the world is limited. These rare congenital anomalies had not been witnessed before in the Gaza Strip, raising an alarm about the relation between congenital birth defects and environmental factors, especially over the last decade.

Keywords: Ophthalmo-acromelic syndrome, spina bifida, hydrocephalus, Gaza Strip

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INTRODUCTION

Mutations in the SMOC1 gene cause OAS. The mutations often result in a non-functional SMOC-1 protein. The loss of SMOC-1 could disrupt growth factor signaling, which would impair the normal development of the skeleton, limbs, and eyes[1].

We noticed that, over the last few years, the incidence of congenital anomalies has increased, especially anomalies related to the heart, and the central nervous and renal systems[2, 3]. Extremely rare cases such as dicephalus parapagus, cloacal extrophy, and prune belly syndrome with vesico-abdominal fistula are being noticed.

Our case was rare; the infant had abnormal fingers and toes, hydrocephalus and lumbar spina bifida. In addition, she was born without eye globes. The case was clinically consistent with OAS[4–6].

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CASE REPORT

Our patient was the fifth child of non-consanguineous parents. She was delivered at full term via normal vaginal delivery. The Apgar scores were seven and nine at one and five minutes, respectively. Her weight at birth was three kilograms. The newborn had anophthalmia, lumbar spina bifida, and macrodactyly. Her feet had two toes each. Her left hand had three fingers, while her right hand had four.

At 30 minutes, the infant was intubated for respiratory distress and connected to a mechanical ventilator. After three days of her birth, she was extubated and started on nasogastric tube feedings. She tolerated the feedings and passed stool. Subsequently, she learned to suck effectively and was fed by a bottle. After 11 days, the infant was operated for lumbar spina bifida. She was discharged at 18 days of age in good general condition and was breathing comfortably in room air. She was hemodynamically stable and able to suck well. However, brain ultrasonography showed severe hydrocephalus. The results of abdominal ultrasonography and echocardiography were normal.

Other Details

Serum electrolytes: Sodium – 137 mmol/l, potassium – 5.7 mmol/l, calcium – 1.14 mmol/l

Kidney function test: Serum urea – 13 mg/dl, creatinine – 1.1 mmol/l

CBC: WBC – 9,600

HGB: 12.3 g/dl

PLT: 358 × 1,000

Figure 1. Her left foot had two toes with macrodactyly

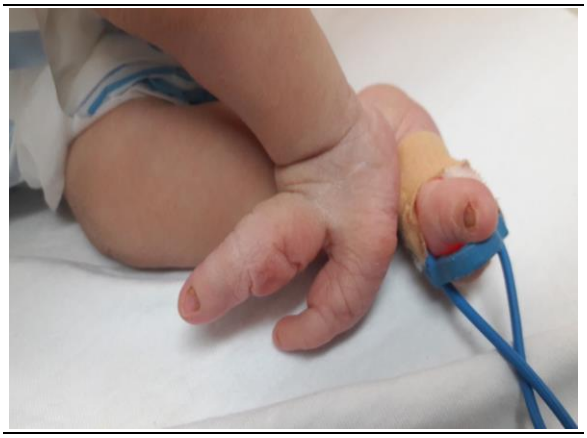


Figure 2. Her right foot had two toes

Figure 3. Her left hand had three fingers with macrodactyly

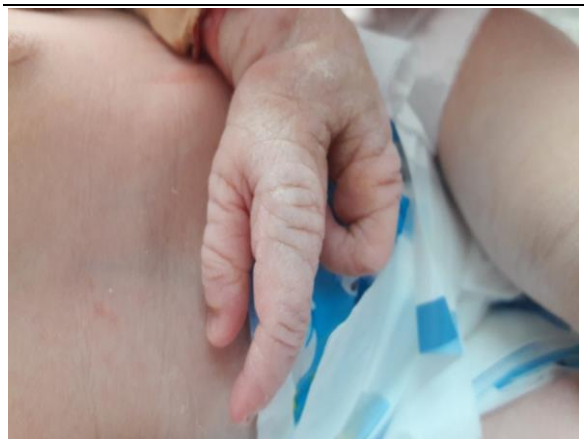


Figure 4. Her right hand had four fingers

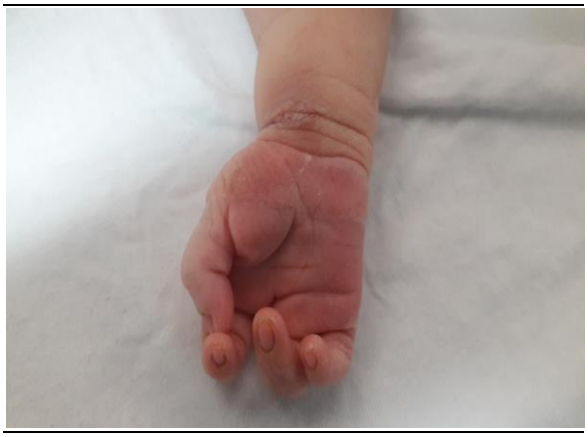
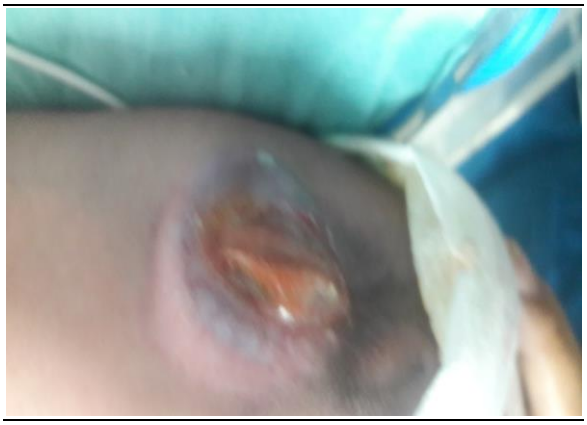


Figure 5. Infant was connected to mechanical ventilator

Figure 6. She had spina bifida



DISCUSSION

The non-consanguineous parents had three daughters and one son before our patient was born; all of them were healthy. Our patient was the fifth child. The family history was negative for such congenital anomalies. The parents were residents of Beit Hanoun in the northern Gaza Strip. This area has been exposed to military attacks involving different weapons.

Studies show a strong correlation between new mutations and environmental pollution caused by heavy metals[2, 3].

CONCLUSION

This was a rare case of OAS with new findings (hydrocephalus and lumbar spina bifida). OAS is an autosomal recessive disease. However, genetic studies were not performed due to limited resources.

Over the last decade, extremely rare congenital anomalies have been reported in the Gaza Strip, especially after three wars. We recommend genetic laboratory tests and more medical research to find the association between birth defects and environmental pollution.

REFERENCES

1. Tekin M, Tutar E, Arsan S, Atay G, Bodurtha J. Ophthalmo-acromelic syndrome: report and review. Am J Med Genet. 2000;90(2):150–4.
2. Naim A, Al Dalies H, El Balawi M, Salem E, Al Meziny K, Al Shawwa R, et al. Birth defects in Gaza: prevalence, types, familiarity and correlation with environmental factors. Int J Environ Res Public Health. 2012;9(5):1,732–47.
3. Manduca P, Naim A, Signoriello S. Specific association of teratogen and toxicant metals in hair of newborns with congenital birth defects or developmentally premature birth in a cohort of couples with documented parental exposure to military attacks: observational study at Al Shifa Hospital, Gaza, Palestine. Int J Environ Res Public Health. 2014;11(5):5,208–23.
4. Abouzeid H, Boisset G, Favez T, Youssef M, Marzouk I, Shakankiry N, et al. Mutations in the SPARC-related modular calcium-binding protein 1 gene, SMOC1, cause Waardenburg anophthalmia syndrome. Am J Hum Genet. 2011;88(1):92–8.
5. Gambhir PS, Gambhir SP, Bankar SM. Ophthalmoacromelic syndrome: two further cases expanding the phenotype. Clin Dysmorphol. 2010;19(2):91–4.
6. Okada I, Hamanoue H, Terada K, Tohma T, Megarbane A, Chouery E, et al. SMOC1 is essential for ocular and limb development in humans and mice. Am J Hum Genet. 2011;88(1):30–41.