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OPHTHALMO-ACROMELIC SYNDROME IN A TURKISH INFANT: CASE REPORT

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H. ÇAKSEN, D. ODABAS, A.F. ONER, M. ABUHANDAN and V. CALEBI

SUMMARY

Ophthalmo-acromelic syndrome (OAS) is an extremely rare autosomal recessive disorder characterised by eye malformations ranging from true anophthalmia to mild microphthalmia and acromelic malformations. In this article, we report a newborn infant with OAS because of its rare presentation. He was the fourth sibling affected in the family. The parents were healthy but there was a close blood relationship between the parents. Physical examination revealed bilateral true anophthalmia and oligodactyly (bilateral four toes) on the feet. He had no other additional abnormalities. We consider that this rare syndrome could be relatively more common in our country because six Turkish cases of OAS have been reported in the English literature to date.

INTRODUCTION

Ophthalmo-acromelic syndrome (OAS) is an extremely rare autosomal recessive disorder characterised by eye malformations ranging from true anophthalmia to mild microphthalmia and acromelic malformations(1-5). Here, we report a newborn infant with ophthalmo-acromelic syndrome.

CASE REPORT

A 14-day-old boy was admitted to our hospital with bilateral anophthalmia. He was the product of a 38 week term uncomplicated gestation and labour. No maternal use of medication or antepartum illnesses were reported. The parents were healthy but there was a close blood relationship between the parents. Our patient was the fifth born in the family. The first three babies (all of them were-girls) born to the family also had bilateral anophthalmia and finger abnormalities, but all had died within the first few months of life from undetermined causes. The fourth gestation resulted in an abortion.

On physical examination, the body measurements were within normal ranges. He had bilateral true anophthalmia and oligodactyly on the feet (Figures 1 and 2). The patient was investigated to exclude other associated abnormalities; however, he had no additional abnormalities. On laboratory investigation, routine blood analysis, renal and liver function tests were normal. The TORCH (toxoplasmosis, rubella, cytomegalovirus, herpes simplex virus) serology was unremarkable. Ultrasonographic and computerized tomography examination of the eyes revealed bilateral absence of the eye bulbs and optic nerves. Unfortunately, we were not able to follow up the patient after discharge from hospital.

Figure 1

Bilateral absence of the eye bulbs is evident



Figure 2

Bilateral oligodactyly on the feet is demonstrated



DISCUSSION

The term anophthalmia refers to congenital absence of one or both eyes. It can be classified into two major types. Primary anophthalmia, in which the ocular primordia never form is very rare and is probably the result of a mutation of genes that are important in the development of the optic vesicle. Secondary anophthalmia is more common and is generally related to infection such as rubella, trauma, vascular events, or toxic/metabolic events (hyper- or hypovitaminosis A) occurring in the early part of the fourth gestational week. In true anophthalmia, neither the optic nerves nor the globes are present(6). Our patient had true anophthalmia and bilateral oligodactyly thus qualifying as OAS. This rare syndrome seems to be common in our country because at least six Turkish cases of OAS have been reported from Turkey between 1996 and 2000(3,5,7,8). We think that this condition is probably related to a high rate (21.1%) of consanguineous marriages in Turkey(9). On the other hand, the presence of a close relationship between the healthy parents, and observation of four siblings with OAS in the family strongly suggest that OAS is an autosomal recessive disorder. Although there is no information about mortality rate of OAS in the literature, it seems that there may be a relationship between mortality rate and the severity of abnormalities because three affected siblings of the family had died earlier.

In OAS, several congenital anomalies such as upper and lower limb abnormalities, split hand, oligodactyly, polydactyly, hypothalamic haemartoma, interruption of the inferior vena cava with azygos and mental retardation have been reported in the

literature(2,3-5,7). Our patient had no additional abnormalities except anophthalmia and oligodactyly.

On account of this case we would like to emphasise that OAS should always be considered in infants with anophthalmia for genetic counseling. In addition we suggest that the adverse effects of consanguineous marriage should be explained to the family in order to reduce the incidence of this severe congenital syndrome.

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