THE CRYPTOPHTHALMOS SYNDROME

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The cryptophthalmos syndrome, first reported by Zehender and Manz in 1872, and described in detail by Francois, is a systemic congenital malformation characterized by cryptophthalmia in association with craniofacial, otorbinolaryngologic, urogenital and extremity abnormalities[1-3]. All of the features of the syndrome may not always appear in the same patient. In this report, a ease with cryptophthalmos syndrome is presented. Clinical and radiologic characteristics, including ultrasonographic and computerized tomographic fmdings, leading to confirmation of the diagnosis are discussed in the view of the literature.

CASE REPORT

A female newborn was immediately brought to neonatal unit of Paediatric Surgery department of Ege University Hospital, Izmir, with multiple congenital anomalies after a normal vaginal delivery. Maternalhistory revealed no significant event during gestational period. On physical examination, the child had anApgar score of nine, birth weight of 3500 gms and length of 60 ems. She had normal vital functions and seemingly active. Her eyes were covered with skin, having no eyebrows, eyelashes and palpebral apertures. Hypertelorism was evident. The nasal bridge was hypoplastie and the aurieles were low set and small in size. The eyeballs were bilaterally palpable, the left one was smaller. There was an osseous defect about 4 x 10 em situated between the right parietal and occipital bones. She had an umbilical cord hernia about 4cm in diameter (Figure 1)

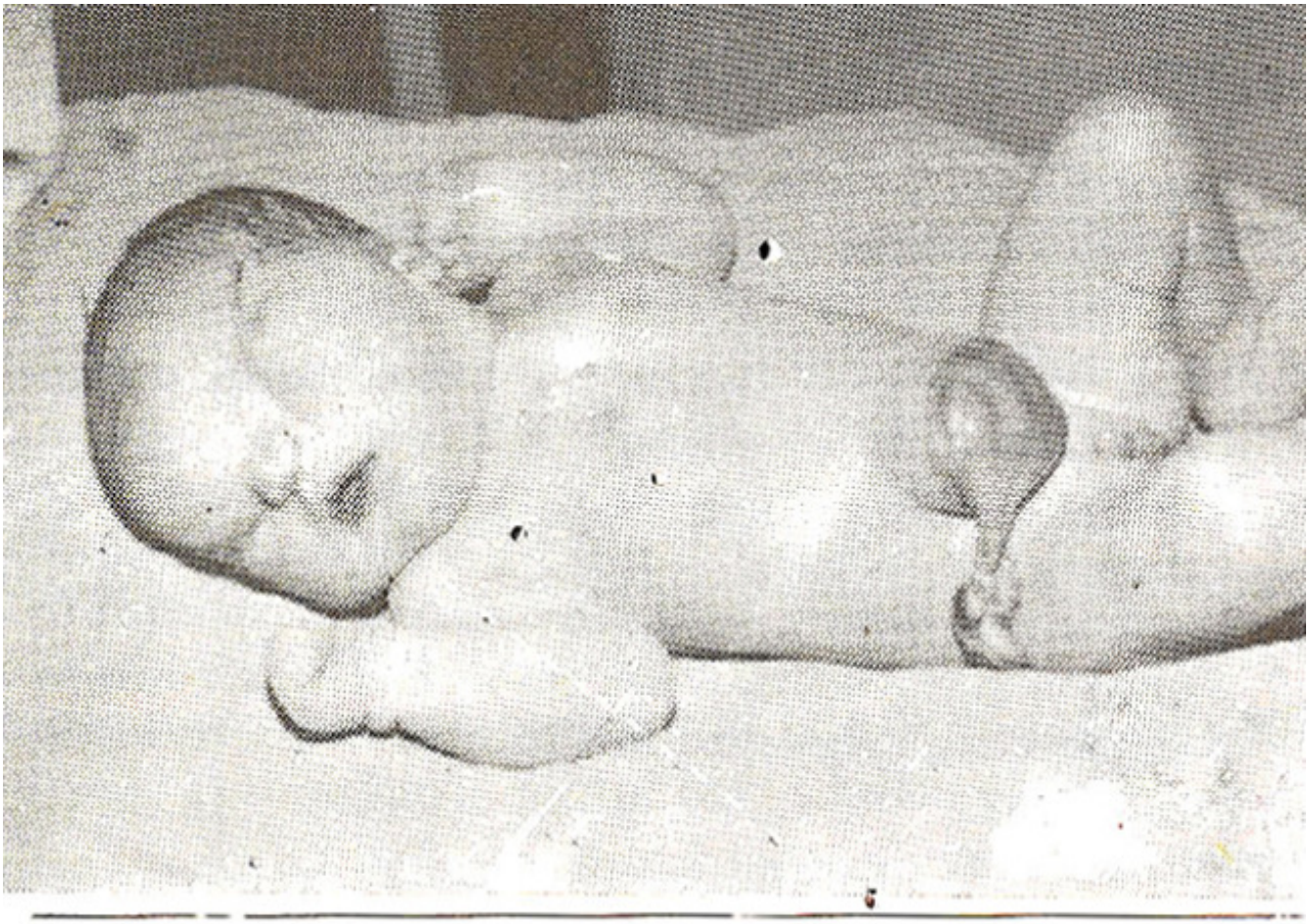

Figure 1. The neonate with cryptophthalmos syndrome.

and a clitoris hypertrophia with normal vagina and anus. There was syndactyly in both of her hands between the 2nd, 3rd, and 4th fingers, and in both of her feet between the 2nd, 3rd, 4th, and 5th toes. No other pathologic findings were noted at the systemic physical examination. In the chromosome analysis, karyotype was 46 XX, sex chromatine was (+) and no genetical defectwas noted. Radiographic evaluation of the vertebral column and extremities revealed no skeletal abnormalities. Orbital ultrasonography confirmed the presence of eyeballs and smallness of the left one. Additionally, both of the lenses were absent. Computerized tomographic scans, performed in paraaxial planes, showed the deformed and elongated eyeballs without having aradiodense lens. Optic nerves and extraocular muscles were normal (Figure 2).

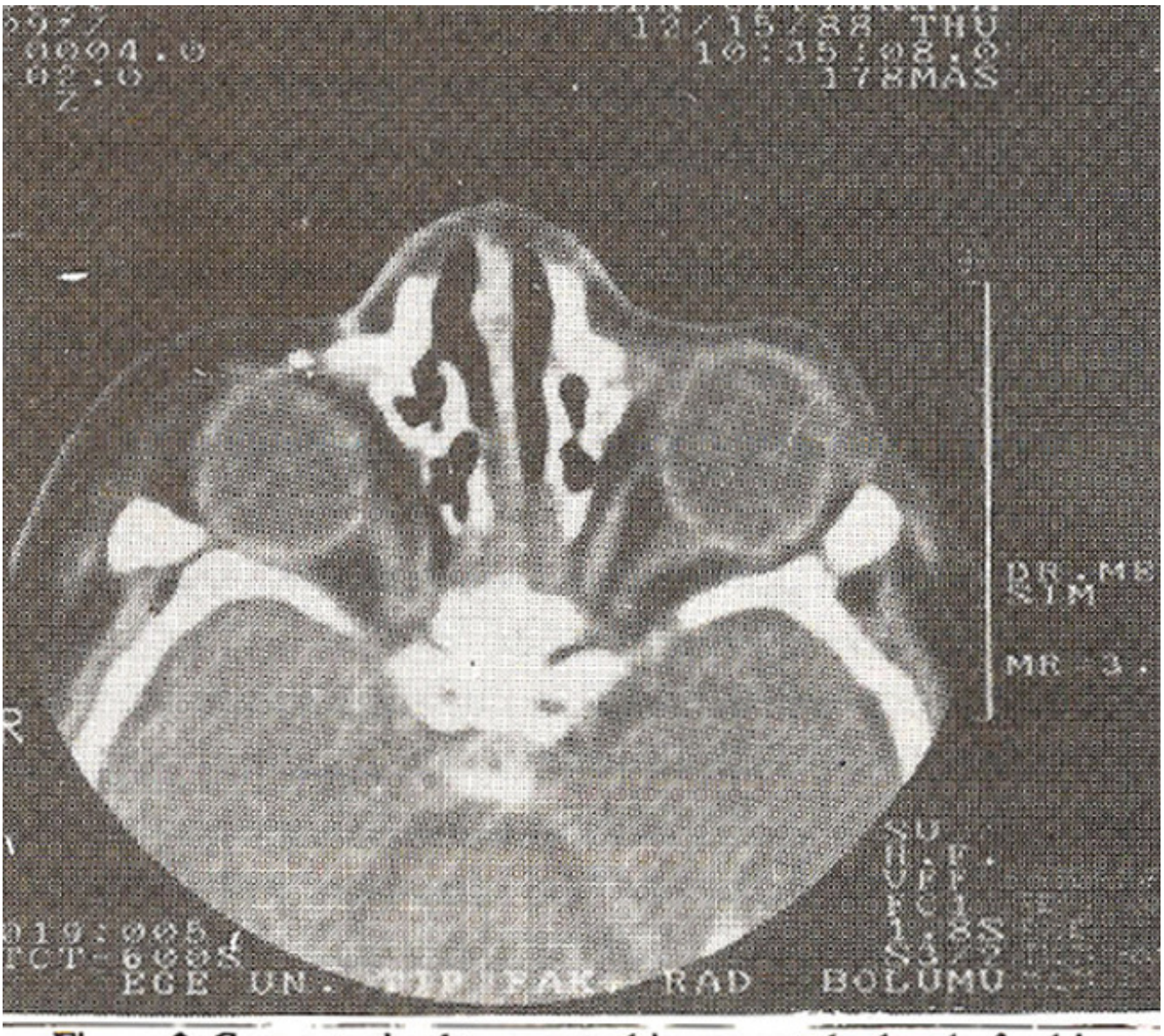

Figure 2. Computerized tomographic scan at the level of orbita.

Left kidneywas absent on the abdominal ultrasonographic evaluation and then in the intravenous urogram. Right kidney and collecting system were normal. The baby underwent a laparotomy for the repair of umbilical hernia and for exploration. She had an incomplete rotation of the intestine with adhesions and peritoneal bands between the duodenum and the right colon. Left kidney was absent and the other abdominal organs were normal. Duodenocolic bands were excised and umbilical hernia was repaired. The repair of syndactyly and the radical operation for the eyeè were deferred to six months of age.

DISCUSSION

Cryptophthalmos syndrome is a systemic mafformation of unknown etiology which is thought to be a genetic disorder with an autosomal recessive mode of inheritance. However, chromosomal defects have never been reported[3,4]. Most often, cryptophthalinia is bilateral. If it is complete, the eyelids are replaced by skin without any palpebral apertures. The eyebrows, eyelashes, meibomian glands, lacrimal glands and lacrimal puncta are generally nonexisting or incompletely developed. The histopathologic

examinations of the skin covering the orbits, have been reported to show metaplasia of the corneal epithelium into the skin[5,6]. In most of the reported cases, there are anomalies in the eye. The incision of the overlying skin, exposes the cavity of globe which contains disorganized tissue including ectatic cornea, vascularized fibrous tissue, an atrophied iris, and if present the lens[5]. Varnek reported colobomatous cysts and microphthalmia in about one fifth of cases[3]. Crarnofacial abnormalities may accompany the ocular fmdings in 25% of cases, including meningoencephalocele, flattening of the frontal area and fusional defects of the face[3]. The osseous defect at the right parietooccipital region of our case was a different manifestation of the cranial anomalies. Ears may be small and low set and external auditory canal may be atretic or stenotic. Malformations of the urogenital system include small penis, hypospadias undescended testis in the male and hypertrophic clitoris with or without vaginal atresia, pseudohermaphroditism in the female and renal agenesia or aplasia[2,3,5]. Ventral hernia is reported in some of the cases, like the umbilical cord hernia in our baby[6]. In the 10% of cases laryngeal atresia or hypoplasia may be seen[3].

Viability is inevitable in these patients. Associated anomalies may be treated easily and successfully, except cryptophthalniia. Bilateral orbital reconstruction including corneal grafting, anterior vitrectomy and lid reconstruction may be performed, but visual deficit may persist due to clouding of the cornea! grafts.

REFERENCES

1. Francois, J. Syndrome malformatin avec cryptophthalmic (Note preliminaire). Ophthalmologica, 1965; 150:215.
2. Konrad, G., Koch, .J.M., Aust, W., Waub, T.N., Forster, M. and Messmer, E. Bilateralervollstrandigerkiyptophthalmus. Klin. MbI. Augenheilk, 1987; 190:121.
3. Varnek, L. Cryptophthalmos, dycephaly, syndactyly and renal aplasia. Acta Ophthalmol., 1978; 56: 302.
4. Sugar, H.S. The cryptophthalmos-syndactyly syndrome. Am. J. Ophthalmol., 1968: 66:897.
5. Levine, RS., Pawers, T., Rosenberg, H.K., Siegel, C.A. and Bilaniuk, L.T. The cryptophthalmos syndrome. MR., 1984; 143:375.
6. Miskinyar, S.A.C., Ossanlon, C. and Argenta, L.C. Ciyptophthalmia; One stage reconstruction. Plast. Reconstr. Surg., 1986; 77:301.