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Cryptophthalmos with an Orbital Cyst and Profound Mental and Motor Retardation

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Cryptophthalmos (hidden eye) or complete ablepharon refers to a congenital anomaly in which lids do not form and skin is continuous over the eye region. This disorder is rare. Approximately 55 cases have been reported in the world literature, with only a few described in the American literature.^{1–7} The syndrome may occur as an isolated disorder, but usually is associated with multiple developmental defects such as nose and ear deformities, cleft palate, cleft lip, ankyloglossia, variations of the hairline, malformations of the teeth, hypertelorism, lacrimal duct defect, laryngeal atresia, renal anomaly, umbilical hernia, meningo-encephalocele, total or partial syndactyly, and incomplete development of the genitalia.

CASE REPORT

This three-year-old white boy, the child of healthy, nonconsanguineous parents with no family history of hereditary disorders or congenital anomalies, weighed 7 lbs. 2 oz. at birth. The child was the product of a full-term, uncomplicated pregnancy with an uneventful delivery. He is the second of three children with completely normal siblings. At birth, the patient was noted to have multiple congenital anomalies including right auditory canal atresia, right microtia with periauricular skin tags, complete cryptophthalmos on the right, partial cryptophthalmos on the left, no palpable globes or tearing bilaterally, a trans-illuminating orbital cyst of the right orbit extending into the glabellar region (Fig. 1), right choanal atresia, ankyloglossia, and a high arched palate without a cleft. Also noted were an enlarged anterior fontanelle and a 2 × 3 cm posterior fontanelle. The head circumference (34 cm) was normal. There was a general hypoplastic appearance of the face, especially on the right. No other abnormal physical findings were observed clinically, except for a simian line of the right palm.

Tests for rubella, cytomegalic inclusion disease, Herpes simplex, and toxoplasmosis were negative. The urine analyses for tyrosine metabolites, keto-acids, mucopolysaccharides, and

amino acids were found to be normal, as were the amino acids in the blood. The VDRL was nonreactive. An intravenous pyelogram demonstrated normal kidneys and urinary tract, and chromosome studies revealed a normal 46, XY male karyotype, confirmed by GTG-banding techniques.

Chest x-rays demonstrated a normal heart size, well ventilated lungs, and slight scoliosis with cartilaginous synchondroses for the posterior arches of the cervical vertebrae. A skull series (4 projections) demonstrated anomalous changes of the calvarium with a markedly hypoplastic frontal bone, and a large prominent sphenoid bone and ridge producing an elevated and poorly developed right orbit. The left orbit was present, but slightly malformed. No frank herniation of the intracranial contents through the bony defects was found. The maxilla was small and hypoplastic, while the mandible also appeared somewhat small. It was noted on the skeletal survey that there was slight bilateral acetabular dysplasia.

At three months of age, agenesis of the corpus callosum was detected by percutaneous ventriculogram. This procedure showed the third ventricle to be increased in size, but no evidence of encephalocele was found. An awake electroencephalogram showed no dominant rhythm, but showed mixed theta rhythm with some superimposed 8–12 cycles per second activity with persistent interhemispheric asynchrony and asymmetry.

This three-year-old child cannot walk, stand, or crawl, and is hyporeflexive. He does not have a social smile and verbal expression remains at the six-month level. The psychological evaluation was done at the age of two years with the Maxfield-Buchholz Scale for the Blind, on which he achieved an MA of five months with an IQ of 23. On the Fairview Self-Help Scale, he achieved an MA of four months with an IQ of 19. Due to the nature of the patient's handicaps (especially blindness), complete standardization testing was difficult and the values achieved were described as tentative.

Audiology tests were done at the age of one year and reflexive responses to high intensity broad band and narrow band (3000 Hz) signals demonstrated normal hearing of the left ear, but no hearing was detected on the right.

Dental records revealed late eruption of the teeth and hypodontia at the age of three years with the absence of one upper central incisor (Fig. 2). The teeth in general are malaligned and irregular in shape.

Surgery was performed to correct the right choanal atresia and to explore the 3.5 cm orbital cyst of the right nasofrontal area. The pathological and microscopic examination of the excised tissue from the cyst demonstrated partially atrophic bundles of skeletal muscle with nonspecific fatty infiltrates surrounding an area of mature fatty tissue with lobular fibrosis and nonspecific focal calcifications in the eye region. There was no inflammation, granulomata, foreign body, neoplasm, or evidence of eye tissue.

DISCUSSION

Cryptophthalmia appears to be an autosomal recessive syndrome^{1,4,8–13} and usually demonstrates four characteristics: cryptophthalmia, dyscephaly (cleft lip and palate, nasal

and ear defects, meningoencephaloceles, etc), syndactyly, and urogenital malformations.¹² Cryptophthalmos is usually bilateral, but five unilateral cases have been reported.¹⁴ The developmental defect may be complete and symmetric, but asymmetry-occurs in about one-third of the cases.¹⁴ The case being reported is bilateral and asymmetric.

Three forms of ocular manifestations are known for cryptophthalmos. These are complete cryptophthalmos, congenital symblepharon, and partial cryptophthalmos, sometimes called hemicryptophthalmos.^{14,15} In complete cryptophthalmos, skin covers the eye totally and attaches to the underlying globe if present. Ocular movements can usually be detected by wrinkling of the skin after shining a bright light into the orbit, producing constriction of the orbicularis oculi muscle. There was no light reflex noted in either orbit of our patient. The eyelashes and eyebrows are usually completely or partially absent.⁵

In congenital symblepharon, the upper eyelid fuses with the globe while the lower eyelid is usually normal. Xerosis of the globe can occur along with keratinization of the cornea.

Partial cryptophthalmos or hemicryptophthalmos occurs in one-fifth of the reported cases.⁵ The eyelids with cilia and an eyebrow are usually present on the lateral side and cryptophthalmos occurs on the medial side of the affected eye. The opposite eye may be normal or malformed.

The right side of our patient demonstrated complete cryptophthalmos and the left side demonstrated partial cryptophthalmos. There was anophthalmos on the right side, as demonstrated by surgery, and extreme microphthalmos or anophthalmos on the left. Extreme microphthalmos or anophthalmos is difficult to differentiate without the use of surgery and microscopic examination.

It is worth noting the profound mental and psychomotor retardation, agenesis of the corpus callosum, and increased size of the third ventricle of our patient as new information for the cryptophthalmos syndrome. Also unique about this case is the occurrence of a large unilateral protruding orbital cyst consisting of skeletal muscle, fatty tissue, and focal calcifications. The growth is not bilateral or confined to the orbital area, as was found in a case reported by Ehlers,¹⁷ but involves the glabellar region as well.

The patient has no genital, renal, or cardiac abnormalities, or syndactyly of the feet or hands, but a simian line was noted on the complete cryptophthalmic side. That these are variable features of the cryptophthalmos syndrome is shown by the report that syndactyly is present in only one-third of the reported cases, while genitourinary anomalies occur in only one-fourth of the cases. Dysplasia of the acetabulum was noted, a finding similar to that reported in a case by Ide and Wollschlager.²

SUMMARY

The cryptophthalmos syndrome generally consists of cryptophthalmia, dyscephaly (cleft lip and palate, nasal and ear defects, meningo-encephaloceles, etc.), syndactyly, and urogenital malformations. This disorder is rare. Approximately 55 cases have been reported, with only a few described in the American literature.

A three-year-old boy with this rare syndrome is described. Major clinical findings include: asymmetric bilateral cryptophthalmos, unilateral orbital cyst of the nasofrontal area, bilateral anophthalmos, right choanal atresia, right microtia, right auditory canal atresia, hypodontia, ankyloglossia, cartilaginous synchondroses of the cervical vertebrae, and bilateral acetabular dysplasia.

Striking features of this case, in addition to the above, were profound mental and motor retardation. Syndactyly and renal anomalies were not observed.

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Fig. 1.
Orbital cyst of the right orbit and glabellar region, deformity of the right external ear.

Fig. 2.
Postoperative view of bilateral asymmetric cryptophthalmos and malformation of the teeth.