Buphthalmos development in adult: case report

Buftalmos em adulto: relato de caso

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ABSTRACT
To report a case of extensive globe enlargement due to secondary glaucoma in a young adult suffering from ocular surface disorders related to hypohidrotic ectodermal dysplasia. To the best of our knowledge, this is the first report of buphthalmos in the adulthood.

Keywords: Hydrophthalmos; Ectodermal dysplasia 1, anhidrotic; Dry eye syndromes; Case report; Humans; Male; Adult

INTRODUCTION
The growth of the human eye occurs more extensively in the postnatal period and is limited to the initial five years\(^1\). Anomalous growth is called buphthalmos and is related to high intraocular pressure in the initial months of life\(^2\).

Gordon et al., in a cross-sectional study including premature, full-term newborns and patients from 2 months to 36 years old, showed the progressive changes during the first years of life. According to their results, the main increase in axial length occurs until the age of 4, then slows to 0.4 mm/year and after 5 years of age, it grows approximately only 1 mm to its final length in adulthood. No significant increase in human eye length is expected after 10-15 years of age\(^3,8\).

The term buphthalmos is used to describe a visible enlargement of the globe at birth or soon thereafter, mostly due to congenital glaucoma. The name buphthalmos comes from the Greek meaning “ox-eyed” and its first formal mention is uncertain and could have been used to highlight clinical diagnoses made by the simple inspection of a large globe. Historically, it was just after the 19th century with the invention of the ophthalmoscope and the tonometer that the etiology of the buphthalmos was related to glaucoma and its distinction of the normally sized protruding globe and other forms eye enlargement were consistently made\(^3\).

We report a case of extensive globe enlargement due to secondary glaucoma in a young adult suffering from ocular surface disorders related to hypohidrotic ectodermal dysplasia. To the best of our knowledge, this is the first report of buphthalmos in adulthood.

CASE PRESENTATION
A 17-year-old caucasian male patient was referred for evaluation of painful ocular surface disorders and uncontrollable and progressive secondary glaucoma. He had been diagnosed with hypohidrotic ectodermal dysplasia in childhood, based on sparse, fine hair, brow and eyelashes hypotrichosis, poor tolerance to heat due to low ability to sweat. At birth his eyes were normal and he developed normal human eye length is expected after 10-15 years of age\(^3,8\).

DISCUSSION
Ectodermal dysplasia (ED) is a congenital syndrome characterized in general terms by sparse hair, severe oligodontia, missing or scanty eye brows, lashes and reduced sweating. It is considered a

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large and complex group of genetic disorders defined by the abnormal development of two or more structures of the ectodermal layer. Nearly 200 different conditions have been described as ED, however, to the best of our knowledge, no previous association with glaucoma or uncontrollable eye growth has been reported.\(^{6,7}\)

Ocular surface manifestations of ED include reduction of eyebrows and lashes, lid keratinization, recurrent epithelial defect, trichiasis, superficial and deep corneal vascularization, limbal deficiency, meibomian glands dysfunction and dry eye.\(^{8,9}\)

Our patient has suffered from ocular surface dysfunction related to ED and developed glaucoma and cataracts secondary to chronic corticosteroid use. We hypothesize that there was a close relation between ED, high IOP and OS globe length growth in this case. The absence of buphthalmos in OD, in which effective IOP control was obtained following trabeculectomy, is an additional evidence of the association of globe length growth and increased IOP in ED patients.

The present work describes a case of ED with challenging management problems that culminated with a unique event of buphthalmos in adulthood.

REFERENCES


Figure 1. A) External appearance at first examination, loss of eye brow and lashes, depressed nasal bridge, epithelial corneal defect in the right eye and buphthalmos at left. B) External appearance after 6 years, showing the progression of the buphthalmos of the left globe. C and D) A-scan ultrasound images of both eyes showing an axial length of approximately 25 mm of the right eye and more than 36 mm of the left.