Short communication

Juvenile glaucoma and optic disc pit with macular detachment in Klinefelter's syndrome

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Abstract

Case report: A 34-year-old man affected by Klinefelter syndrome, with loss of vision in his left eye (LE), ocular hypertension with increased cupping in both eyes, and optic disc pit with serous macular detachment in the LE. Optical coherence tomography showed a macular detachment with a double-layer detachment, consisting of both an inner layer separation and an outer layer detachment. The outer layer detachment did not seem to communicate with the optic disc.

Discussion: Klinefelter syndrome has been associated with diffuse choroidal atrophy and colobomas of the iris and choroid. Our patient showed bilateral juvenile glaucoma and unilateral congenital optic disc pit. This association has not been previously reported in the bibliography.

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Introduction

The Klinefelter syndrome, 47 XXY, is the most frequent chromosomal aberration among men, with a frequency of 1:500 births and the most prevalent cause of male infertility. It is associated to low levels of testosterone, sterility due to azoospermia, low bone density, large height, disproportionate bodies with long legs and short torso, gynecomastia, small testicles, feminoid distribution of bodily hair and language alterations. There is a substantial variation in the clinical presentation which can make diagnostic difficult. Testosterone treatment corrects the symptoms derived from the deficiency of androgens but has no effect on fertility. The Klinefelter syndrome has been associated to various ocular diseases such as diffuse choroidal atrophy and iris and choroids colobomae.

A case report is presented which associates the Klinefelter syndrome to juvenile glaucoma and optic pit with macular serous detachment. In the reviewed references said association has not been found.

Case report

Male, 34, diagnosed with Klinefelter syndrome visited the emergency section due to visual acuity loss in the LE with 2 days evolution. Visual equity in the right eye (RE) was of 1 and in LE of 0.2. The anterior pole did not exhibit alterations and the angle was open, grade IV 360°, poorly pigmented. Intraocular pressure (IOP) of the RE was of 34 mmHg and 30 mmHg in the LE. Central corneal thickness in the RE was of 516 μm and in LE of 520 μm. The funduscopy assessment revealed a marked increase of papillary cupping in both eyes with a cup/vertical disc quotient of 0.8. The LE exhibited an oval shaped depression corresponding to a congenital pit of the optic nerve in the temporal sector with a peripapillary atrophy area associated to macular serous detachment (Figs. 1 and 2). The study of the retina nervous fiber layer with optic coherence tomography (OCT) (Stratus OCT Carl Zeiss Ophthalmic Systems, Inc.) revealed a significant thinning in both eyes, more marked in the RE (Fig. 3). However, it must be taken into account that the actual thickness of the left optic nerve fiber layer could be increased due to the macular serous detachment. Computerized campimetry (Humphrey 24-2) revealed a superior arch-shaped defect in the RE and a central defect in the LE due to the maculopathy (Fig. 4). Typically, fluorescein angiography showed initial pit hypofluorescence (Fig. 5). OCT over the macular revealed the separation of the internal retina layers in communication with the pit without connection with said pit. It also revealed a lamellar macular hole in the external layers of the retina (Fig. 6).

Discussion

The association between the Klinefelter syndrome, chronic open angle glaucoma and optic nerve pit with macular serous detachment has not been described.

Chronic juvenile glaucoma performs similarly to adult open angle primary glaucoma, following the same treatment algorithm. It is characterized by high IOP caused by increased resistance to the exit of the aqueous humor at the level of the trabecular mesh and a generally advanced optic neuropathy at diagnostic time. Juvenile glaucoma is transmitted through dominant autosomic inheritance with reduced penetrance and the gene encoding the mycillin protein (MYOC) has been identified as responsible for this disease. It is not known whether the association with the Klinefelter syndrome is casual or if both share the same etiopathogenic basis.

Congenital optic nerve pits consist in a colobomatous defect of the optic papilla. Colobomae at the level of the iris and the choroids have already been described in the Klinefelter syndrome and therefore the association of said syndrome with the papillary pit could share the same etiopathogeny. Optic nerve pits are generally unilateral, with visual acuity and visual fields normal although some associated campimetric defects have been described. Maculopathy appears in up to 2/3 of cases with a mean age of 30 years. Multiple etiopathogenic theories have been described to explain the origin of the subretinal liquid in macular detachment associated to the papillary pit, including vitreous origin, choroidal exudation and fistulization of the cerebro-spinal fluid from...
the subarachnoid space through the papillary pit. The initial injury which corresponds to a cystic separation of the internal layers, while a posterior degeneration of the external layers could give rise to a genuine serous detachment with separation of the photoreceptor layer from the pigment epithelium layer. The persistence of the lesion causes the degeneration of the external retina layers and the opening of partial thickness macular holes, as in this case report, or even total thickness macular holes. This detachment of external layers is not usually complicated with the optic nerve as shown by the OCT in the present case (Fig. 6A).

Spontaneous resolution of the maculopathy has been described in 25% of untreated cases, although the perpetuation thereof involves irreversible changes on the macula. Three months is the maximum period of time recommended to wait before taking action to achieve macular reapplication. Some authors have proposed photocoagulation of the temporal retina adjacent to the papilla with the intention of sealing the passage of fluid from the pit to the macula. However, this treatment is not always effective but could be considered as an initial therapeutic option. Surgical treatment is indicated if the visual acuity worsens or the macular detachment exceeds

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Fig. 3 – Optic coherence tomography: defects in the retinal fiber layer of both eyes.

Fig. 4 – Visual field. (A) Upper arch-shaped defect in the right eye. (B) Central defect in the left eye.

Fig. 5 – Fluorescein angiography: initial hypo-fluorescence corresponding to the optic nerve pit.

Fig. 6 – Optic coherence tomography. (A) Image showing the optic nerve pit and the communication between the internal retina layers and said pit. The neurosensory detachment (arrow) does not communicate with the optic nerve. (B) Image of the macular serous detachment with lamellar hole (arrow) in the external layers of the retina.
3 months. Vitrectomy is performed with the intention of eliminating the vitreous traction on the pit, considering that this traction is a significant pathogenic factor in the associated macular detachment.5

Conflict of interests

No conflict of interests has been declared by the authors.

REFERENCES


