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CASE SERIES OF EYE DISORDERS ASSOCIATED WITH CONGENITAL EYE SYNDROMES
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CASE SERIES OF EYE DISORDERS ASSOCIATED WITH CONGENITAL EYE SYNDROMES

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ABSTRACT

***Background*: Congenital diseases are sometimes incompatible with life. Others are, but sometimes at the cost of suffering for the child and family. These abnormalities often have a rich symptomatology and interest several specialties. Ocular signs rarely help in prenatal diagnosis. After birth, the ophthalmologist may contribute to early diagnosis of these diseases generally serious.**
***Objective*: To identify ophthalmological signs of some congenital diseases.**
***Design*: A descriptive and transversal study.**
***Subjects*: Four children were seen between January 2012 and December 2014 for ophthalmological damages due to congenital disease.**
***Results*: Ophthalmological lesions observed were: lens subluxation with high myopia due to Marfan's syndrome, retinitis pigmentosa and obesity due to Bardet-Biedl's syndrome, cryptophthalmia due to Fraser's syndrome, dermoid cyst in a context of Goldenhar's syndrome.**
***Conclusion*: The ophthalmologist can be the first to move towards the congenital disease diagnosis.**

INTRODUCTION

Congenital diseases are hereditary in their majority, with one or more known genetic mutations. The mutation can be transmitted by the parents, or so-called "mutation de novo " when there is not a proven family inheritance (1, 2). These diseases may also be the consequence of the teratogenic effects of embryofetopathy or toxicity of some drugs (3). Congenital diseases are sometimes incompatible with life. Others are, but sometimes at the cost of suffering for the child and family. These abnormalities often have a rich symptomatology and interest several specialties. Ophthalmological signs may contribute to early diagnosis of these diseases generally serious (4). This survey aims to identify the ophthalmological signs of congenital diseases diagnosed in four children seen in the ophthalmology department of the University Hospital of Brazzaville (UHB), namely: Marfan's syndrome, Bardet-Biedl's syndrome, Fraser's syndrome and Goldenhar syndrome.

MATERIALS AND METHODS

It was a series of four children aged 6 months to 16 years, seen for ophthalmological damages due to a congenital disease. It was a descriptive and transversal study, conducted between January 2012 and December 2014 (3 years) in the ophthalmology department of the UHB. The data was collected through a survey sheet. As part of this study, each child had been seen once.

RESULTS

Case one: A 8 years old girl was seen for bilateral visual blur. At the age of 3 she's already suffering from myopia, -5 diopters (D). A new pair of glasses was prescribed every 9 months due to a rapid progression of this myopia which reached the value of-12 D when she is 7 years. Furthermore, parents noted the exceptional flexibility of the joints of their child, as well as her large size compared to other children of

the same age in the family. On admission the review noted, on both sides, high myopia- 18D, inferonasal subluxation of the lens (Figure 1), dolichostenomelia, thinness (Body Mass Index = 12.75), arachnodactyly, thoracodorsal scoliosis. The diagnosis of Marfan syndrome had retained.

Figure 1

Cardiovascular examination was normal. Surgery (lensectomy) was carried out on both sides. The post operative period was favorable (visual acuity= 10/10 on both sides with a correction of - 1D after one month). Cardiovascular and ophthalmological monitoring had been recommended for life.

Case two: A 16 year old girl was seen in consultation for a bilateral blurred vision lasting for nearly 2 years. His review had noted on both sides: a bilateral blindness (counting fingers at 20 cm), retinitis pigmentosa. Also included: obesity (Body Mass Index = 34.32), vaginal atresia, surgical scars on the outer side of each little finger witness of polydactyly (Figure 2), mental retardation. The diagnosis of Bardet-Biedl syndrome was retained.

Figure 2

A multi disciplinary care has been recommended (low vision unit, speech therapy care, renal assessment and vaginoplasty).

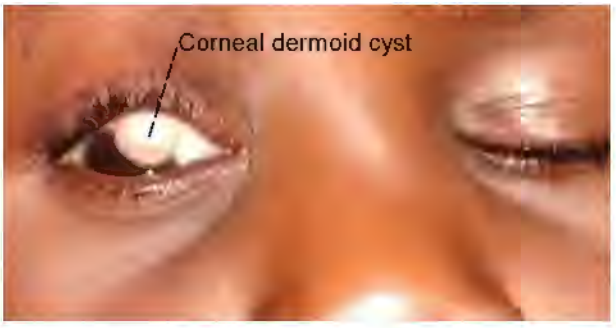
Case three: A male child, aged 6 months was referred for a right orbital congenital malformation. His review noted (Figure 3): incomplete cryptophtalmia (presence of a draft of the upper eyelid, without palpebral fissure and without eyeball), surgical scar treatment of labial cleft palate, bilateral syndactyly (ring and little fingers were merged). Left eye examination and the general examination were normal. The diagnosis of Fraser syndrome was retained.

Figure 3

Plasty of the face had been programmed.

Case four: A congenital tumor affected the right eye of a 4 year old boy. The lesion was typically hemispheric, covered with pink skin, located at the supero nasal part of the cornea (Figure 4).

Figure 4



This tumor is responsible for a significant corneal astigmatism at the origin of major amblyopia (visual acuity limited to counting fingers at 30 cm). There were no other major abnormalities in the clinical examination but a single skin tag was observed at the homolateral preauricular area without any visible cutaneous fistula next to it. This additional sign and the pathological examination of a biopsy specimen, allowed us to establish the diagnosis of a minor form of Goldenhar's syndrome. An ablation of the tumor with corneal transplant was performed. An orthoptic rehabilitation has been recommended.

DISCUSSION

Case one: Marfan's syndrome is a rare genetic disease; with an autosomal dominant transmission. However in a third of cases are spontaneous mutations, not inherited. It is due to connective tissue injury, in relation to an anomaly of fibrillin, which is a component of elastin. This disease affects all organs of the body, with very variable degrees of clinical manifestations. The most affected organs are eyes, the cardiovascular system and the skeleton (4). The most frequent ocular damage are: subluxation of the lens, cataract, myopia, glaucoma and retinal detachment. These eye lesions will benefit from standard treatments, often with satisfactory functional results. The great size, arachnodactyly, scoliosis and dolichostenomelia are the main skeletal damage. Aneurisms and dissections of aorta make gravity of Marfan's syndrome. Other serious but rare complications can occur: heart failure in mitral insufficiency, heart rhythm disorders. Apart from any valve disease, electrocardiogram often shows nonspecific abnormalities. Original rhythm of ventricular disorders can cause sudden death. Beta-blockers cushion the impact of systolic flow on the weakened media of the aorta. They reduce the progression of aortic dilatation and may reduce the risk of complications although this is not proven (4, 5).

Case two: Bardet-Biedl's syndrome is a rare genetic disease. It is autosomal recessive. Several genes responsible for this disease have recently been localized. Its diagnosis is clinical and based on the combination of two major criteria and three minor criteria (6, 7). The major criteria are: retinitis pigmentosa, obesity, polydactyly, delay learning, sexual abnormalities (hypogonadism, bicornuate uterus, vaginal atresia ...). The minor criteria are: delay in speech, cataracts, strabismus, brachydactyly, syndactyly, diabetes insipidus, diabetes mellitus, abnormalities of dentition, congenital heart disease. The forms without renal impairment are better prognosis; mortality from this syndrome is essentially due to nephropathy. Laurence-Moon-Biedl-Bardet's syndrome is often confused with Bardet-Biedl syndrome, but strictly speaking it associates spastic paraplegia and polydactyly is absent (8).

Case three: Fraser's syndrome is a rare genetic disorder transmitted in an autosomal recessive mode. Its diagnosis is clinical based on the presence of two major criteria (cryptophtalmia, syndactyly, genital anomalies) and one minor criteria (ear abnormalities, abnormalities of the nose, larynx anomalies and / or palate, skeletal abnormalities, umbilical hernia, renal agenesis, mental retardation), or one major and four minor criteria (9, 10). The only cryptophtalmie without syndactyly may be a differential diagnosis with two other birth defects. Blepharophimosis syndrome which is a rare congenital orbito frontal eyelid malformation, autosomal dominant, affecting both sexes. It combines a malformation of the bony orbit (small and depressed, with a flattened upper orbital rim), a malformation of the eye (epicanthal folds, a telecanthus, and ptosis due to the levator muscle hypotrophy associated with short eyelids), characteristics eyebrows (hypertrichosics shaped), an inconstant ectropion of the outer part of the eyelids. The palpebral coloboma, which are defined by a deficit in the eyelid margin. Many pathophysiological hypotheses are mentioned, such as defects of contiguous embryonic bud or mesenchymal tissues. They can be defined according to their size, their seat, their severity, their number and presence of associated abnormalities in soft tissue and skeleton, which will be systematically on the same axis (10).

Case four: The Goldenhar syndrome or atrio-eye-vertebral dysplasia is a rare genetic disease, autosomal recessive. It primarily affects the ocular apparatus, the hearing system and the skeleton in addition to mental retardation. The symptoms have variable degree. In addition to auditory, ocular and skeletal abnormalities, some patients may have serious visceral lesions. The main ocular abnormalities are corneal dermoid cyst, the eyelid coloboma and microphthalmia (11). Other anomalies are very diverse but the most encountered are represented by the absence of the pinna, the absence of the ear canal, hypoplasia of the mandibular bone, heart defects, kidney and genital malformations (11, 12).

In conclusion, the diagnosis of congenital diseases is generally multidisciplinary, including Obstetricians, Pediatricians and Ophthalmologists when there are associated ocular abnormalities. Recognition of eye damage can help early diagnosis and thus improve vital and / or functional prognosis.

REFERENCES

1. Li, Y. and Lin, H. Progress in screening and treatment of common congenital eye diseases. *Eye Sci.* 2013; **28**: 157- 162.
2. Vachha, B. A. and Robson, C. D. Imaging of Pediatric Orbital Diseases. *Neuroimaging. Clin. N. Am.* 2015; **25**: 477- 501.
3. Chabrolle, J. P., Bensouda, B., Bruel, H., Simon, A., Poinsot, J., Ickowicz, V., *et al.* Metopic craniosynostosis, probable effect of intrauterine exposure to maternal valproate treatment. *Arch. Pediatr.* 2001; **8**: 1333- 1336
4. Kwun, Y., Kim, S. J., Lee, J., *et al.* Disease-specific Growth Charts of Marfan Syndrome Patients in Korea. *J. Korean Med. Sci.* 2015; **30**: 911-916.
5. von Kodolitsch, Y., De Backer, J., Schüler, H., Bannas, P., Behzadi, C., Bernhardt, A. M., *et al.* Perspectives on the revised Ghent criteria for the diagnosis of Marfan syndrome. *Appl. Clin. Genet.* 2015; **16**: 137- 155.

6. Chennen, K., Scerbo, M. J., Dollfus, H., Poch, O. and Marion, V. Bardet-Biedl syndrome: cilia and obesity - from genes to integrative approaches. *Med. Sci. (Paris)*. 2014; **30**: 1034-1039.
7. M'hamdi, O., Ouertani, I. and Chaabouni-Bouhamed, H. Update on the genetics of bardet-biedl syndrome. *Mol. Syndromol*. 2014; **5**: 51-16.
8. Laurence, J. Z. and Moon, R. C. Four cases of retinitis pigmentosaoccuring in the same family and accompanied by general imperfections of development. *Ophtalmic. Rev*. 1866; **2**: 32-41.
9. Touré, A., Diomandé, I. A., Nouraly, H., *et al*. Bilateral cryptophthalmos in Fraser syndrome: Case report and review of the literature. *J. Fr. Ophtalmol*. 2015; **38**: 97- 100.
10. Tran, A. Q., Lee, B. W., Alameddine, R. M. *et al*. Reconstruction of Unilateral Incomplete Cryptophthalmos in Fraser Syndrome. *Ophthal. Plast. Reconstr. Surg*. 2015.
11. Baugh, A. D., Wooten, W., Chapman, B., Drake, A. F. and Vaughn, B. V. Sleep characteristics in Goldenhar Syndrome. *Int. J. Pediatr. Otorhinolaryngol*. 2015; **79**: 356-358.
12. Puvabanditsin, S., February, M., Francois, L., Garrow, E., Bruno, C. and Mehta, R. 7q21.11 Microdeletion in a Neonate With Goldenhar Syndrome: Case Report and a Literature Review. *Cleft. Palate. Craniofac. J*. 2015.