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Ophthalmological manifestations in children with Down Syndrome 21: about 65 cases

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ABSTRACT

Ocular manifestations are frequent in trisomy 21 and are dominated by refractive disorders. Management should be early with specialized and systematic pediatric ophthalmic examinations. The majority of these anomalies are treatable to ensure better social integration. We report a study describing these manifestations in 65 children with Down syndrome 21 followed in the university hospital of Marrakech.

KEY WORDS: Trisomy 21, refractive disorders, amblyopia, strabismus, keratoconus.

INTRODUCTION

Down syndrome child 21 can present a series of visual problems that often go unnoticed. This is all the more regrettable as their treatment usually leads to a noticeable improvement in vision and quality of life. The most common problems are related to a refractive disorder, which impairs vision and does not encourage The aim of this study is to determine the frequency and characteristics of the ophthalmological manifestations encountered in children with Down Syndrome.

MATERIAL AND METHODS

This is a retrospective study of 65 children with Down syndrome 21, recruited in pediatric ophthalmology consultation at the Mohamed VI University Hospital Center in Marrakech over a period of 4 years from January 2012 to November 2015. Diagnosis of the syndrome Down has been established from clinical and genetic data. All children underwent a complete ophthalmologic examination, including cycloplegic refraction, visual behavioral analysis, visual acuity measurement for older children and, in the absence of significant mental retardation, anterior segment examination. posterior and measurement of ocular tone. An orthoptic assessment and a general pediatric assessment were also established.

RESULT AND DISCUSSION

The average age of the patients was 5 years (range from 6 months to 15 years). Male predominance was noted in 60% of cases (sex ratio = 1.5). In most cases, consultation was part of the systematic evaluation of children with Down Syndrome 21 (60%), in 30% of cases this consultation was motivated by strabismus. The ophthalmological manifestations encountered in our series (Figure I) were dominated by refractive disorders (Figure II): 58.7% hyperopia, 37% astigmatism and 26%

myopia.A medium astigmatism (<1.5 D) was more frequently found in our patients. Children older than 10 years had more astigmatism compared to other age groups (Figure III). Strong myopia (> 6 D) was observed in 9 cases (13.84%). Other ocular manifestations were represented by strabismus noted in 38% of cases, amblyopia in 10.76% and nystagmus in 7.69% of cases. Blepharo-conjunctivitis was common (27.7%). Congenital cataract was found in 9.2% of cases, while congenital glaucoma was noted in 3% of children. The extraocular abnormalities noted in our patients were dominated by cardiac malformations (7 cases), followed by testicular ectopia (2 cases), congenital dislocation of the hip (1 case), congenital megacolon (1 case) and deafness. (1 case). The management consisted of an optical correction of the refractive anomalies associated with the treatment of amblyopia, a medical treatment for children with blepharoconjunctivitis. The surgical treatment consisted of cataract surgery (6 cases), trabeculectomy (2cas), dacryorhinocystomy (2 cases), transfixing corneal transplantation (1cas) (figure IV). For keratocaine, we performed a gas injection into the anterior chamber (Figure V)), in addition to medical treatment (hypotonisants and corticosteroids).

Table 1 : the ophthalmological manifestations encountered in our patients.

Ophthalmological event	Number	Percentage
astigmatism	24	36,92%
hyperopia	38	58,46%
Myopia	17 2	26,15% 3%
keratoconus	25	38,45%
Strabismus	7 5	10,76% 7,69%
amblyopia	6 2	9,2% 3,07 %
nystagmus	4	6,1%
Congenital cataract	18	27,69%

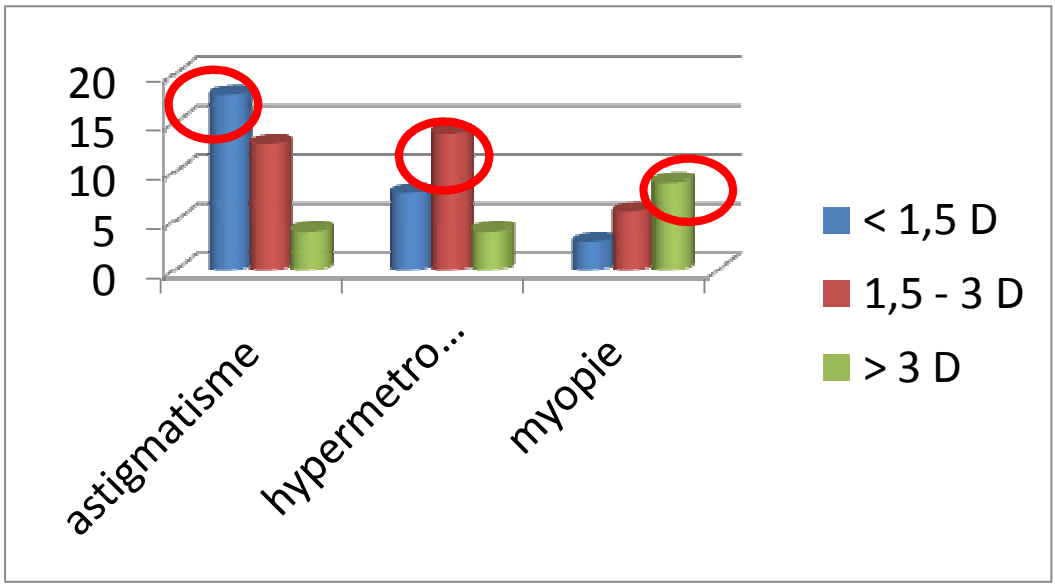

Figure I: refractive status of our patients.

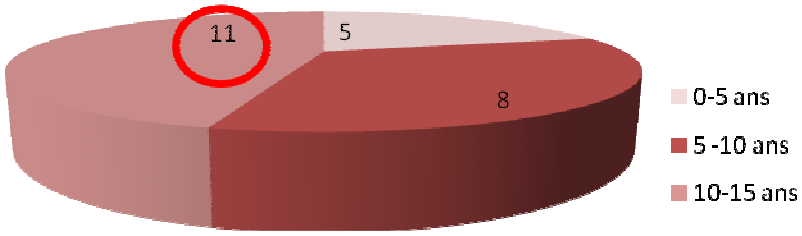

Figure II: astigmatism as a function of age.

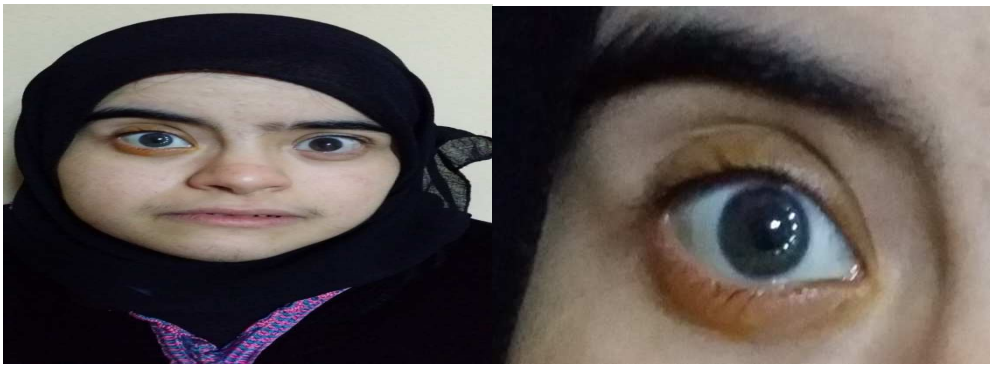

Figure III: keratoplasty transfixing for keratoconus

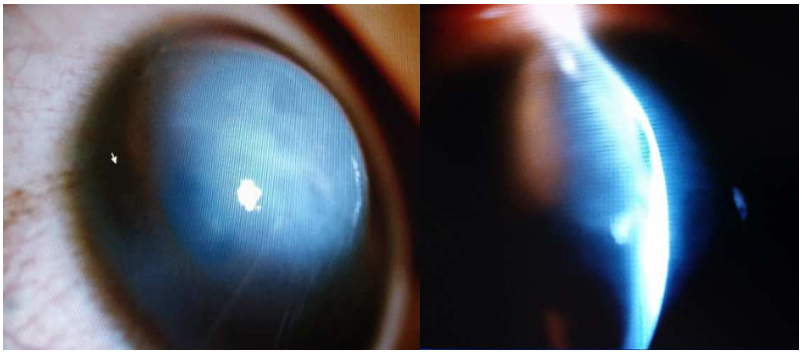

Figure V: Gas injection into the anterior chamber for acute keratoconus.

Discussion

Trisomy 21 is a genetic anomaly that affects the twenty-first pair of chromosomes.The diagnosis is discussed clinically, confirmed by the karyotype. Children with Down Syndrome present the same eye diseases as the general population with the same risks for vision and the same treatments available. Nevertheless, some diseases are more frequent in these people and therefore require special attention, in order to detect and manage them in time. Studies show that more than 90% of children with Down syndrome 21 who have been seen by an ophthalmologist have at least one ocular problem [1,2].

The most common problems are related to a refractive disorder. According to the series, 70 - 75% of people with trisomy 21 have an ocular refractive disorder that is distributed almost equally between myopia, hyperopia and astigmatism [3]. Strabismus is most often present very early, before age. 6 months, must be diagnosed early. It is sometimes secondary to an ocular refraction disorder and appears as a common pathology since 15 to 20 times more frequent than in the normal population: ± 35% against 1 to 2% [4-6]. Comparing our results with those of In the literature, ametropia remains the most frequent anomaly in trisomy 21 with percentages varying between 40 and 90%, followed by strabismus (Figure I). The prevalence of these abnormalities increases with age, unlike children with normal development who become more emmetropic with age [12]. The evaluation of visual acuity and the management of amblyopia remain difficult given the particular contact with these children. Monitoring of visual function is essentially based on the measurement of the angle of deflection and the refractions under cycloplegic treatment every 6 months [13].

Keratoconus also appears more frequent, since it affects 2 to 3 people per 1000 in the general population but 5 to 15% of people with trisomy 21. Unfortunately the discovery is quite often made in advanced stages with consequent deep-sightedness associated [14].

It has also been described the existence of corneal opacities that can interfere with vision. Congenital cataract and glaucoma were less common but had potentially serious implications for later vision [15]. The frequency of congenital cataract in our series is close to that observed in the English, Egyptian and Nigerian series [3].

Children with trisomy 21 also appear to be more prone to acute infectious diseases such as conjunctivitis or blepharitis. Even if the treatment is the same as in the normal population, it is none the less present in a chronic form in 30 to 40% of cases [3]. Malformations of the nasolacrimal system have also been reported, represented essentially by obstruction of the nasolacrimal duct [16].

Iris abnormalities include anterior stromal hypoplasia and Brushfield stains. Although found in the

general population, these irises are more common and characteristic of Down syndrome 35 to 78% of newborns. Brushfield spots are easily identifiable without magnification, although iris staining may be the basis of some differences [17]. The prevalence of nystagmus in these children ranges from 9% to 30% [18,19].

Other eye abnormalities reported in Down syndrome include retinal dystrophy with optic atrophy, choroidal sclerosis, retinoblastoma and uveitis [7,20]. Retinal detachment has also been described [21].

Management should be comprehensive for both ophthalmic and general malformations. The presence of cardiovascular abnormalities aggravates the vital prognosis and complicates the management of the patient during general anesthesia.

CONCLUSION

Children with trisomy 21 have characteristic eye manifestations and associated problems and must undergo periodic ophthalmic examinations. Down syndrome eye abnormalities are, for the most part, treatable and a significant visual loss should rarely occur in these patients. The diagnosis and early treatment of these problems are essential to obtain optimal vision as an adult and will allow the patient with Down syndrome to function at his highest potential, which will facilitate his social integration. .

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