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Research Article

Diagnosis of Rare Pediatric Diseases by Retcam Imaging

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Abstract

Purpose: To highlight the findings of rare pediatric lesions that were imaged by Retcam, that added a very important tool to diagnose hereditary or sporadic blinding diseases in infants.

Setting: The Ophthalmic Diagnostic Laser Unit, Kasr Al-Ainy Hospitals, Cairo University.

Design: Cross sectional, non-randomized, observational case series.

Subjects and Methods: Colored fundus (CF) findings and fluorescein angiography (FA) of 10 Egyptian infants and children referred for Retcam imaging to diagnose and confirm a retinal pathology causing visual disability.

Results: Two cases were diagnosed as optic disc swelling, one proved to be papilledema due to premature closure of cranial sutures in Down syndrome and the other was diagnosed as Para infectious optic neuritis. Two cases were diagnosed as Aicardi syndrome showing typical retinal lesions. Two cases were diagnosed as congenital toxoplasmosis, one of them showed active lesions at retinal periphery. One case was diagnosed as retinal hamartomas of Tuberous Sclerosis, and another case was diagnosed as combined hamartoma of retina and retinal pigment epithelium (RPE). The last two cases were pigmentary retinopathy, one of them showed a well developed picture of retinitis pigmentosa, although aged 4 years.

Conclusion: Retcam imaging shouldn't be restricted to the screening of retinopathy of prematurity (ROP) or follow-up of conservative treatment for retinoblastoma, but its wide field imaging can diagnose multiple mysterious causes of pediatric visual disability.

Keywords: Retcam; Pediatric; Retinal Disorders; Visual Disability

Introduction

Indirect ophthalmoscopy for fundus examination for infants and children has proved its validity over the past years, however, interpretation and accurate diagnosis needs great experience and assistance. The field of view using the 20 diopters lens (60° field of view) or even the 28 diopters lens is small (69° field of view) [1], maybe full of reflections, with absence of documentation and fluorescein angiography if needed. The Retcam imaging system provides high-quality photographs that can be obtained by trained non-physician personnel and evaluated by a remote expert. It has been shown to have high reliability and accuracy in detecting referral-warranted Retinopathy of prematurity (ROP). Additionally, the method is generally well received by parents and is highly cost-effective [2]. The ability of capturing movies and selecting the best photos after the end of the examination, in addition to the documentation for late or remote consultancy add to its value in examining infants. Therefore Retcam imaging may be also used to differentiate many confusing and mimicking diseases in the pediatric age group, especially when using fluorescein angiography, like Coat's disease, retinoblastoma when appearing as small peripheral masses and other hamartomas.

The aim of this study is to highlight the findings of rare pediatric lesions that were imaged by Retcam, which added a very important tool to diagnose hereditary or sporadic blinding diseases in infants.

Patients and Methods

This is a cross-sectional, non-randomized observational case series, carried out on a cohort of 10 Egyptian infants and children referred for Retcam imaging to diagnose and confirm a retinal pathology causing visual disability. The study was performed during

the time period between August 2015 and August, 2017. The examination was performed at the Ophthalmic Diagnostic Laser Unit at Kasr Al-Ainy Hospitals of Cairo University. Kasr Al-Ainy Hospitals represent a tertiary referral center for most of the governorates in Egypt. The study was approved by the Research Ethics Committee of the Faculty of Medicine, Cairo University. Data collection conformed to all local laws and was compliant with the principles of the Declaration of Helsinki.

All parents received a thorough explanation of the study design, imaging procedure, anesthesia and possible risks of anesthesia and fluorescein injection if used and provided their written consents.

Patients included in this study; were infants referred to diagnose the cause of visual disability noticed by the parents, thorough history of the labor circumstances, gestational age, birth weight, consanguinity, and all associated mishaps of pregnancy or medications were recorded. Cases referred for Retinopathy of Prematurity (ROP) staging or follow-up of retinoblastoma were excluded, as the aim of the work was to highlight unusual findings in this age group.

Methodology in details

All patients underwent Retcam imaging using RetCam 3 Wide-Field Digital Imaging System (Clarity Medical Systems, Inc., Pleasanton, CA) using the wide field 120° lens. The parents were instructed that the children should be fasting for at least 4 hours prior to the examination, pupillary dilation was done using tropicamide 1%, Light general anesthesia was given to the child using Laryngeal mask airways or endotracheal tube if needed. Fluorescein Sodium 10% was used if needed using only I cc for IV injection. A wire speculum was inserted in the eye after application of topical anesthesia (Benoxinate Hydrochloride 0.4%). Hydroxypropyl Methylcellulose coupling fluid was used before the application of the camera hand piece on the globe. Video capturing was performed for colored and fluorescein angiography photos for both eyes. Choice of best photos from the videos was then performed, saved and analyzed.

Results

Case 1

Female infant aged 8 months having Down's syndrome with bilateral mild ptosis and was referred for Retcam imaging due to suspicious fundus lesion. Diagnosis of papilledema and left ischemic optic disc due to premature closure of fontanelles and cranial sutures (Craniosynostosis) was proved by radiology (Figure 1).

Case 2

A female full term infant aged 18 months presented with bilateral optic disc swelling. FA proved the optic disc edema and leakage of active optic neuritis which is very rare in this age (Figure 2).

Figure 1: CF and FA of case 1 showing bilateral optic disc edema more evident in the right eye, with areas of choroidal non perfusion and ischemic optic disc in very late phases in the left eye.

Figure 2: CF and FA of bilateral optic neuritis.

Case 3

Female child aged 18 months presented with nystagmus and muscular spasms. FA wasn't needed for the diagnosis. The fundus shows bilateral multiple retinal lacunae diagnosing Aicardi syndrome.

Figure 3: CF of case 2 showing multiple whitish lesions known as retinal lacunaes and small peripapillary choroidal colobomas.

Case 4

Female child aged 24 months presented with nystagmus and also infantile seizures. The fundus shows right retinal lacunae and small peripapillary colobomas and left morning glory appearance diagnosing another case of Aicardi syndrome.

Figure 4: CF of case 3 showing right small optic disc coloboma with retinal lacuna extending nasally and left large optic disc and choroidal coloboma (Morning Glory Syndrome).

Case 5

Male child aged 1 year presenting with right exotropia and poor following response. Retcam imaging showed right irregular macular lesion that proved to be scarred lesion with no leakage in FA and well circumscribed suspicious of congenital toxoplasmosis that was proved by lab tests.

Figure 5: CF and FA of the right eye showing circumscribed macular scar.

Case 6

Female patient aged 3 years presenting also with right esotropia and poor vision. Retcam imaging showed large circumscribed macular lesion. Retcam FA with wide field lens 120° showed hyperfluorescent RPE window defect of macular scar suggestive of old lesion of toxoplasmosis, however the peripheral area shows active leaking lesions of active toxoplasmosis.

Figure 6: CF and FA of the right eye showing macular circumscribed scar and peripheral active leaking lesions.

Case 7

5 months old boy presented with right esotropia. CF shows multiple whitish lesions. The differential diagnosis was retinoblastoma versus astrocytic hamartoma.

The masses were translucent, overlying the vessels showing early hyperfluorescence and late staining in FA with no evidence of feeder vessels as in retinoblastoma. Thus astrocytic hamartoma was diagnosed and proved by MRI showing paraventricular subependymal astrocytic nodules and Confetti skin lesions of Tuberous sclerosis.

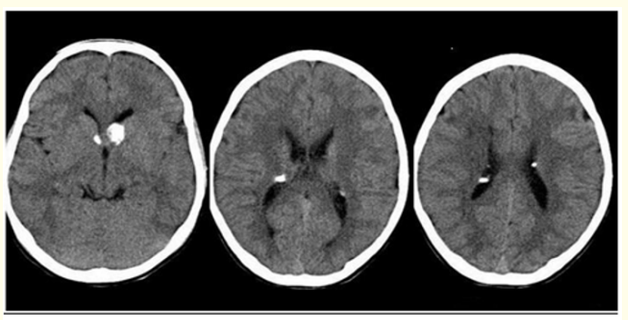

Figure 7: CF and FA showing multiple translucent hyperfluorescent masses overlying the vessels with late staining. The skin shows Confetti lesions and MRI shows paraventricular lesions.

Case 8

Male child aged 2.5 years old presented with left esotropia, CF showed macular graysh elevated lesion with temporal retinal ridge and dot hges. FA showed macular hyperfluorescent lesion with microaneurysms and dilated capillaries, the peripheral temporal retina shows avascular retina similar to stage 2 ROP. The diagnosis was combined retinal and retinal pigment epithelium (RPE) hamartoma.

Figure 8: CF showing a macular grayish lesion that shows dilated capillaries and mild leakage in FA and peripheral retinal avascularity.

Case 9

A 4 years old female presented with poor vision. CF shows pigmentary retinopathy with macular involvement. FA showed attenuated vessels, macular and peripheral hyperfluorescent RPE defects and pigmented specules, diagnosing atypical retinitis pigmentosa, that was proved by ERG.

Figure 9: CF and FA of bilateral a variant of retinitis pigmentosa.

Case 10

A 2 years old female showing bilateral pigmentary retinopathy (Salt and pepper appearance).

Figure 10: CF and FA of pigmentary retinopathy.

Discussion

Fundus examination of newly born babies, infants and children is usually difficult without anesthesia or sedation. Many abnormalities can be seen which maybe rare or having a modified appearance adding difficulties for an appropriate diagnosis.

Documentation of findings and addition of fluorescein angiography for this age group provide a much better way to improve our diagnosis and maybe better management and rehabilitation.

Wide field Retcam imaging was introduced for screening of Retinopathy of Prematurity (ROP) in premature units. Digital photography had a sensitivity of 100% and specificity of 97.5% in detecting prethreshold and threshold ROP. Positive-predictive value of digital photography was 67% and negative-predictive value was 100%. Screening and management of ROP using RetCam imaging did not fail to detect prethreshold or threshold disease when images could be obtained [3]. However many authors reported its use in detection and follow-up for other diseases like screening retinal hemorrhages in healthy newborns [4]. Intraoperative imaging with wide-field photography and echography are complementary approaches commonly employed to aid in the diagnosis and management of retinoblastoma, especially with the evolution of new lines of treatment from enucleation and external beam radiation therapy (EBRT) to other globe-salvaging therapies, including chemoreduction with focal consolidation [5] and, more recently, superselective intraarterial chemotherapy [6].

In our case series many infants were referred from pediatricians or young ophthalmologists for the diagnosis of retinal or optic nerve lesions causing visual disability.

The aim of study is to demonstrate rare and unusual findings in some cases examined by Retcam imaging, that required further investigations and highlighted uncommon pathology.

In cases 1 and 2, there was unusual optic disc edema to be detected at this young age (8 months and 18 months, respectively). Papilledema due to premature closure of cranial sutures and fontanelles (Craniosynostosis) was the diagnosis of case 1 (Figure 1), which is a rare finding that was further proved by CT scans, as Down's syndrome is commonly associated with delayed closure of cranial sutures and fontanelles. There is no known association between craniosynostosis and Down syndrome, despite the latter condition is having well-described craniofacial anomalies. Two patients with trisomy 21 Down syndrome and single-suture craniosynostosis were described by Siu., *et al* [7].

The other case was diagnosed as Parainfectious optic neuritis (Figure 2). This condition is usually bilateral in pediatric age group. The interval from febrile illness to symptom onset was reported to be six days in the pediatric group and 19.5 days in the adult group. The authors postulated that the diagnosis of optic neuritis days to weeks after the presumed systemic infection supports the theory that parainfectious optic neuritis is due to an immulologic-inflammatory reaction [8]. The diagnosis was made due to bilaterality, history of febrile illness and recovery.

The other two cases (3 and 4) were diagnosed as Aicardi syndrome. Aicardi., *et al*. first described this condition, now named after him, in 1965. It is a rare neuro-developmental disorder originally characterized by the triad of; infantile spasms, partial or total agenesis of the corpus callosum, typical lacuna-shaped chorioretinal lesions [9]. The chorioretinal lacunae consist of well-circumscribed, full-thickness defects limited to the retinal pigment epithelium (RPE) and choroid, with an intact overlying retina that may appear histologically abnormal. These lesions are most commonly found around the optic nerve head and the posterior pole and typically decrease in size towards the peripheral fundus (Figure 3). Previous reports have noted increased pigmentation or fading of

lacunae over time. Unilateral chorioretinal lacunae do not rule out the diagnosis of Aicardi syndrome in the presence of the clinical picture of the syndrome 9. Other ophthalmic features include optic nerve coloboma or hypoplasia and microphthalmia. Other features have been reported more sporadically, including optic nerve aplasia, increased rate of excavated disc anomalies, morning glory abnormality (figure 4), nystagmus, sixth cranial nerve palsy, persistent pupillary membrane, iris cyst, iris coloboma, aniridia, peripheral retinal dysplasia, glial tissue extending from the optic disc [9].

Cases 5 and 6 were diagnosed as congenital toxoplasmosis, which was proved by lab tests. The unusual findings were the presence of well circumscribed scarred unilateral retinochoroidal lesion in such a young age (1 year in case 5) and peripheral active lesions in case 6. The incidence of peripheral lesions was proved to be higher in black races than white races [10].

Cases 7 and 8 described retinal hamartomas and highlighted how to be differentiated by FA and other ancillary tests as MRI from retinoblastoma or mimicking diseases. Two of the major criteria for the diagnosis of Tuberous Sclerosis were found in case 7 as retinal hamartomas and subependymal nodules and one minor manifestation were the Confetti skin lesions (Figure 7), which proved the diagnosis according to the UPDATED DIAGNOSTIC CRITERIA FOR TUBEROUS SCLEROSIS COMPLEX [11].

Combined hamartoma of retina and RPE was the diagnosis of case 8 and was established by FA as described by Schachat., *et al* [12]. The FA showed early hypoflourescence due to blockage in the region of hyperpigmentation. In the arterial-venous phase, microaneurysms and a fine network of abnormal dilated capillaries with leakage were observed. The peripheral lesions were characterized on fluorescein angiography by straightening of the vessels and relative avascularity. A report by Helbig., *et al*. [13] discussed a case with significant areas of capillary non-perfusion that resulted in preretinal neovascularization peripheral to the hamartoma, suggesting that significant retinal ischemia can occur.

Finally case 9 showed retinitis pigmentosa manifested so early as full blown picture of retinitis pigmentosa, although the age was young (4 years), Tsujikawa., *et al*. [14], stated that the average age when patients were diagnosed with RP was 35.1 years, and the median age was 36.5 years. The onset ratio straightly increased with age until 65 years, so the onset ratio was relatively low at young ages.

Case 10 was comparatively diagnosed as pigmentary retinopathy with no manifestations of retinitis pigmentosa, which may develop later into a phenotype of retinitis pigmentosa.

Conclusion

In conclusion this case series highlighted unusual rare retinal pathologies in pediatric age and demonstrated the other clinical manifestations that can help in the diagnosis.

Conflict of Interest

The authors declare no conflict of interest in the material discussed in this study.

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Financial Disclosure

The authors declare no financial interest in the material discussed in this study.

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