· Case report ·

A new eye syndrome of cataract, dwarf, polydactyly associated with genu valgum?

Xiu-Qin Zhao[1], Xin Zhao[2], Wen-Zhong Yang[1], Jie Yang[2], Dennis Lam[3]

[1] Department of Ophthalmology of CPLA-234 Hospital, Chaoyang 122000, Liaoning Province, China
[2] Department of Ophthalmology, the People's Hospital of Guangxi Zhuang Autonomous Region, Nanning 530021, Guangxi Zhuang Autonomous Region, China
[3] Department of Ophthalmology & Visual Sciences, the Chinese University of Hong Kong, Hong Kong Eye Hospital, Kowloon District, Hong Kong, China
Correspondence to: Dennis Lam. Director of Department of Ophthalmology & Visual Sciences, the Chinese University of Hong Kong, Hong Kong, China. Chief of Shantou University Chinese University of Hong Kong Joint SIEC. dennislam@cuhk. edu. hk
Received:2010-05-26 Accepted:2010-07-23

Abstract

• A male patient, 39 years old, presented with symptoms of cataract, dwarf, progeria, polydactyly and genu valgum, after the extracapusular cataract extraction and intraocular lens implantation on both eyes, the visual acuity was improved and his fundus examination was normal. As the syndrome is different from any eye syndrome we have ever known, so it maybe a new one.
• KEYWORDS: cataract; dwarf; polydactyly associated with genu valgum; new eye syndrome
DOI:10.3969/j. issn. 1672-5123. 2010. 08. 005

Zhao XQ, Zhao X, Yang WZ, Yang J, Lam D. A new eye syndrome of cataract, dwarf, polydactyly associated with genu valgum? *Int J Ophthalmol (Guoji Yanke Zazhi)* 2010;10(8):1469-1470

INTRODUCTION

We report a case with symptoms of cataract, dwarf, progeria, polydactyly and genu valgum, as the syndrome didn't meet any eye syndrome we have ever known, so it maybe a new one.

CASE REPORT

A male patient, 39 years old, had blurred vision in both eyes for more than 10 years, and the condition got worse for 3 months. For his poor economic condition, he didn't come to hospital until the disease seriously affected his life. He had normal mental state, old looking, normal intelligence level, and the ability to handle his normal life. Physical examination: he had hypermicrosoma, 1.59 meters in height (Figure 1), and he looked like 50 years old though he was 39. He had irregular tooth alignment with protruding front teeth (Figure 2). His four limbs were short and associated with polydactyly (Figure 3). He had the symptoms of the skeletal system: spinal fusion and posterior scoliosis (Figure 4). He was confirmed without consanguineous marriage. Ophthalmology examination: the visual acuity of his right eye was light perception, and it couldn't be corrected. Light position check was all normal. Color vision was normal. The visual acuity of his left eye was counting fingers /20cm, and it couldn't be corrected, either. Light position and color vision were normal. Cornea was transparent in both eyes. Anterior chamber length was 2.5CT. Pupil size was 2.5mm, round with sensitive light reflection. Both lenses were gray with complete opacification (Figure 2). Vitreous and retina couldn't be checked. B-type ultrasound didn't find any change in vitreous. This patient received extracapusular cataract extraction and intraocular lens (ECCE + IOL) implantation on both eyes. After surgery, visual acuity of right eye was 0.5; left eye 0.6 (could not be corrected both). Fundus examination was normal. He was discharged from hospital after one week, and followed up for 4 months. No complication was observed. We considered this man got an amblyopia.

DISCUSSION

Clinical features (1) This patient was 39 years old, the cause of his cataract was unknown. His mother also had low visual acuity, so we consider that it is hereditary, but it is hard to prove because of his mother's death. (2) As he had the symptoms of four short limbs associated with polydactyly, spinal fusion and posterior scoliosis, irregular tooth alignment with protruding front teeth, and genu valgum, so we concluded that he had multiple bone deformity. (3) He had a short stature, 1.59 meters in height. (4) He had an old looking when he was 39 years old, which indicated that he had progeria.

Difference between this syndrome and others The syndrome in this patient had the combined manifestations of cataract, progeria, dwarf and bone deformity, we consider it an eye-related syndrome. When we compared it with those syndromes we have known, there are some similarities, but they are not exactly the same. (1) Difference from Lanzieri syndrome: the cause of Lanzieri syndrome cause is still not clear. Although it also has dwarf, irregular tooth alignment, it still has congenital missing of fibula, tarsal, plantar unit, and hirsutism[1]. These were not found in this case. In eyeball, the ocular manifestations of Lanzieri syndrome have not only cataract, but also the defect of iris, choroid and optic nerve. These also did not appear in our case. Polydactyly was observed in this case while it never appears in Lanzieri syndrome. (2) Difference from Gansslen syndrome: the symptoms of Gansslen syndrome include dwarf, polydactyly

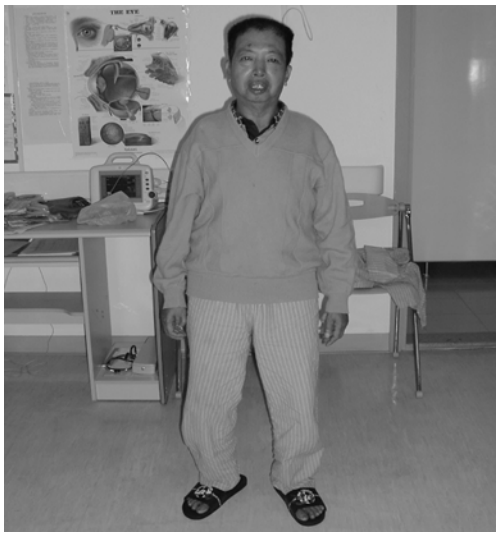

Figure 1 Dwarf.

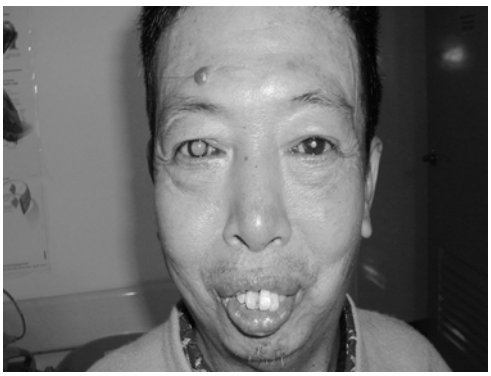

Figure 2 Progeria and irregular tooth alignment.

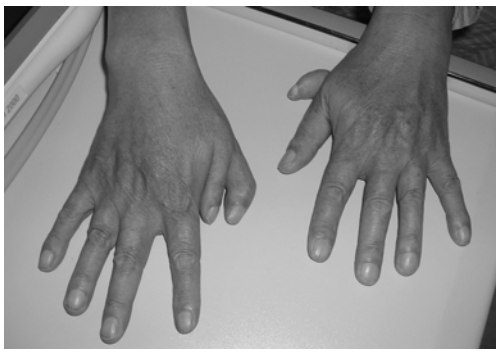

Figure 3 Four limbs were short with polydactyly.

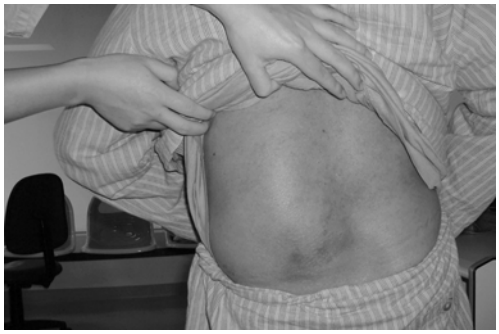

Figure 4 Spinal fusion and spinal scoliosis.

and cataract, etc, while its cataract is congenital[1]. Besides, Gansslen syndrome has familial hemolytic jaundice, hepatosplenomegaly and oxycephaly. It still has small eyeball, the defect of iris and choroid, pupil displacement. These did not show in our case. (3) Difference from Ellis-van Creveld syndrome: Ellis-van Creveld syndrome is autosomal recessive, occurred in 25% descendants of consanguineous marriage[2, 3]. Its ocular manifestations include cataract, iris defect, and the physical symptoms include dwarf, polydactyly, genu valgum and low mental development. In our case, iris defect was not found, and the patient's intelligence level was normal.

Is it a new syndrome? Our case is different from those syndromes we have mentioned, as progeria never appears in Lanzieri syndrome, Gansslen syndrome or Ellis-van Creveld syndrome. So we regard it as a new syndrome type, combined with dwarf, progeria, polydactyly, genu valgum and cataract. After the ECCE + IOL implantation on both eyes, the visual acuity of right eye was improved to 0.5, and left eye 0.6. No change was observed in fundus.

REFERENCES

1 Zhao XQ, Zhang J. Eye Syndrome. Beijing: China Radio and TV Press 1995: 105, 154

2 Ellis RWB, van Creveld S. A syndrome characterized by polydactyly, chondro-dysplasia and congenital morbuscordis. *Arch Dis Child* 1940; 15: 65

3 Lin SC, Zhao XQ. Common Eye Syndrome. Beijing: People's Medical Publishing House 2008: 180-181

白内障、侏儒、多指及膝外翻,一种新的综合征?

赵秀琴[1],赵 昕[2],杨文忠[1],杨 捷[2],林顺潮[3]

(作者单位:[1]122000 中国辽宁省朝阳市,中国人民解放军 234 医院眼科;[2]530021 中国广西壮族自治区南宁市,广西壮族自治区人民医院眼科;[3]中国香港特别行政区九龙区,香港中文大学眼科及视觉科学学系)

作者简介:赵秀琴,女,主任医师,研究方向:青光眼、眼科综合征及眼部整形。

通讯作者:林顺潮,香港中文大学眼科及视觉科学学系主任,汕头大学·香港中文大学联合汕头国际眼科中心院长,国际眼科委员会及国际眼科理事会成员,2008 年世界眼科大会主席,世界青光眼协会委员会委员,亚太区眼科学会秘书长及 CEO,《国际眼科杂志》名誉总编,发表国际论文 500 余篇. dennislam@cuhk.edu.hk

摘要

患者,男,39 岁,具有白内障和侏儒、早老症、多指及膝外翻等症状,双眼行白内障摘除+人工晶状体植入术后视力有所提高。眼底检查未见异常改变,因这些症状不符合以往已知的一些眼病综合征,我们推测其为一种新的眼科综合征。

关键词:白内障; 侏儒; 多指及膝外翻; 新的眼科综合征