CASE REPORT

High Myopia Associated with Triple X Syndrome

Tomo Nishi and Nahoko Ogata

Department of Ophthalmology, Nara Medical University, Nara, Japan

ABSTRACT

We report our findings in a 3-year-old girl who was suspected of having triple X syndrome because she was taller than +4.35 standard deviations for her age. She also had high myopia. Optical coherence tomography (OCT) showed that her retinas were thin, the lenses were subluxated, and the axial length was elongated. Our findings indicate that for a female child with tall stature, there should be thorough evaluations for endocrinological disorders, overgrowth syndromes, connective tissue disorders, and genetic disorders. If there are also behavioral issues, this may lead to consideration of 47 XXX in a female or 47 XXY in a male infant. The 47 XXX syndrome is a potential and neglected cause for tall stature and requires a high index of suspicion.

ARTICLE HISTORY

Received 22 February 2016
Revised 12 March 2016
Accepted 15 March 2016

KEYWORDS

Children; high myopia; tall stature; triple X syndrome

Introduction

Triple X syndrome is a sex chromosome anomaly in women who have an extra X chromosome. The most common physical features include tall stature, epicanthal folds, hypotonia, and clinodactyly.^{1,2} In Japan, the incidence of triple X syndrome is approximately 1 in 1000 female births.³ There have been reports on the signs and symptoms of triple X syndrome; however, high myopia and ectopia lentis have not been reported in triple X syndrome. We report a case diagnosed triple X syndrome by karyotyping who had a tall stature and also high myopia.

Case Report

A 3-year-old girl was noted to have visual impairments during a routine checkup and was brought to our hospital for an evaluation of her vision. She was born by normal delivery at a gestational age of 38 weeks with a birth weight of 2732 g. Her visual acuity was 20/300 in the right eye and 20/500 in the left eye. The intraocular pressure was 10 mm Hg OD and 12 mm Hg OS. In dim illumination, the diameters of both pupils were 3 mm. Both pupils were round and reacted normally to direct light, but a relative afferent pupillary defect was present in the left eye. The

refractive error (spherical equivalent) after cycloplegia with atropine was −16.00 dioptres in the right eye and −20.00 dioptres in the left eye. Both of the anterior segments were normal, but both lenses were subluxated into the vitreous cavity. Ophthalmoscopy showed myopic chorioretinal atrophy and tilted discs (Figure 1A, 1B).

Optical coherence tomography (OCT) showed a thinning of the retina, with the central foveal thickness of 212 µm in the right eye and 206 µm in the left eye, compared with a mean of 220 µm in normal eyes in our clinic (Figure 2A, 2B). The axial length was 24.92 mm in the right eye and 26.44 mm in the left eye, which are markedly longer than the 21.72 mm reported for 20 normal children of mean age of 6.7 years.⁴

She was 108 cm tall, which was +4.35 standard deviations taller than the normal height for Japanese girls at this age. Because of her excess height, she consulted a paediatrician to rule out any systemic abnormalities. Her facial features were normal, and she did not have abnormal epicanthic folds. Her Japanese Newborn screen was normal, heart examinations were normal, and no murmur was detected. Her intelligence was within normal limits for her age. No hormonal abnormalities or pituitary gland lesions were found. She was

Figure 1. Fundus photographs of both eyes of a patient with triple X syndrome. The right eye (1A) and the left eye (1B) show retinal atrophy and a tilted disc.

Figure 2. Optical coherence tomographic (OCT) image of the same patient with the triple X syndrome. 2A: Horizontal scan of the right eye. 2B: Horizontal scan of the left eye. 2C: Vertical scan of the right eye. 2D: Vertical scan of the left eye. Both retinas are thinner and the eyes have a posterior staphyloma.

Figure 3. Result of the chromosome examination. Circle indicates the extra X chromosome.

diagnosed with the triple X syndrome because karyotyping showed a 47XXX karyotype (Figure 3).

She was prescribed −7.0 dioptres glasses for the right eye and −8.0 dioptres for the left eye. One year later, the best-corrected visual acuity (BCVA) of the right eye improved to 20/200 and that of the left eye to 20/400.

Discussion

Triple X syndrome is a rare sex chromosome anomaly. Severe medical problems can be associated with triple X syndrome, e.g., delay of growth, congenital hydrocephalus, premature ovarian failure, frame abnormalities, and renal dysplasia.¹ Our patient did not have any of these signs except for the tall stature. At present, no ocular abnormalities have been reported for individuals with the triple X syndrome.

Our patient was diagnosed with triple X syndrome after an examination of her chromosomes by karyotyping, but other signs and symptoms, except the height, were not present. Bilateral lens subluxation and tall stature can be associated with Marfan syndrome. However, her paediatric examinations were normal because her Japanese Newborn screen was normal, heart examinations were normal, and no murmur was detected. X-ray photographs of her hand and spine were normal, so Marfan syndrome was ruled out. Although she was only 3 years old, she had unusually high myopia, lens subluxation, and an elongation of the axial length. However, a quantification of the retinal layers showed that the retina was significantly thinner than that for children at this age.

Search of MEDLINE with keywords “triple X syndrome” and “ophthalmological anomalies” did not extract any publications. However, Klinefelter syndrome, which is considered to be a relative of triple X syndrome, has been reported to be associated with high myopia.^{5–7} There are case reports

of a 28-year-old⁵ and an 8-year-old⁶ with Klinefelter syndrome with high myopia whose vision were improved by optical corrections.

Because of the rarity of the triple X syndrome, we have been able to study only one patient. However, we present these findings to alert the ophthalmological community that an unusually tall individual with high myopia may have the triple X syndrome and should be karyotyped. In addition, we recommend that a complete ophthalmological examination including OCT be performed on such individuals so that any retinochoroidal abnormalities can be found and corrected.

Declaration of Interest

The authors report no conflicts of interest. The authors alone are responsible for the content and writing of the paper.

References

- [1] Tartaglia NR, Howell S, Sutherland A, Wilson R, Wilson L. A review of trisomy X (47,XXX). *Orphanet J Rare Dis* 2010;5:8. doi: [10.1186/1750-1172-5-8](https://doi.org/10.1186/1750-1172-5-8).
- [2] Liebezeit BU, Rohrer TR, Singer H, Doerr HG. Tall Stature as presenting symptom in a girl with triple x syndrome. *J Pediatr Endocrinol Metabol* 2003;16:233–235.
- [3] Maeda T, Ohno M, Matsunobu A, Yoshihara K, Yabe N. A cytogenetic survey of 14835 consecutive live births. *Jinrui Idengaku Zasshi* 1991;36:117–129.
- [4] Nishi T, Ueda T, Hasegawa T, Miyata K, Ogata N. Choroidal thickness in children with hyperopic anisometropic amblyopia. *Br J Ophthalmol* 2014;98:228–232.
- [5] Karampelas M, Gardner J, Holder G, Hardcastle A, Webster A. Retinal dysfunction and high myopia in association with 48,XXYY syndrome. *Doc Ophthalmol* 2013;127:245–247. doi: [10.1007/s10633-013-9406-x](https://doi.org/10.1007/s10633-013-9406-x).
- [6] Qureshi F, Thurairajan G. Case report: 49, XXXXY syndrome and high myopia. *Ophthalmic Physiol Opt* 2007;27:500–501.
- [7] Van Splunder J, Stilma JS, Evenhuis HM. Visual performance in specific syndromes associated with intellectual disability. *Eur J Ophthalmol* 2003;13:566–574.