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**Application of
Diagnosis Using the
Digital-photoscreener
in Chinese Patients
with X-linked ocular
albinism**

Michelle Lee¹ and Xiangming
Guo^{1,2}

¹State Key Laboratory of Ophthalmology,
Zhongshan Ophthalmic Center, Sun Yat-
sen University, Xianlie Road, Guangzhou
510060, China

²Corresponding author: Xiangming
Guo, Pediatric and Genetic Eye Clinic,
Zhongshan Ophthalmic Center, Sun
Yat-sen University, 54 Xianlie Road,
Guangzhou 510060, China

Purpose: Ocular albinism type 1 (OA1) and congenital motor nystagmus (CMN) are two kinds of eye diseases that are easily confused in Chinese patients. The purpose of this study is to evaluate a simple and efficient diagnosis method for OA1 patients using the Digital Photoscreener.

Methods: Sixteen Chinese OA1 patients harboring FRMD7 mutations and 12 Chinese CMN patients harboring FRMD7 mutations participated in this study. The Digital Photoscreener and ophthalmological examination were performed at the Pediatric and Genetic Eye Hospital in the Zhongshan Ophthalmic Center. Standard analytical procedures were used to compare the results obtained from the ophthalmological examination with the results obtained from Photoscreener photographic analysis. Furthermore, 30 Chinese normal controls also took part in as match.

Results: The photographs taken by Digital Photoscreener for all patients with OA1 showed mild transillumination in the iris and abnormal red reflections from the fundus of the eyes. The obvious red color shown in the pupil region made it easy to distinguish OA1 from CMN because the photographs of all patients with CMN and the 30 normal controls did not show red reflections of the fundus.

Conclusions: The Digital Photoscreener greatly increases the ability to screen for a wide range of ocular problems. However, its efficacy in diagnosing OA1 has not been documented. In our study, we found that Digital Photoscreening is an efficient technique for screening OA1 and distinguishing OA1 from CMN in Chinese patients. By using the Photoscreener, we can easily observe that photographs taken of those with OA1 differ significantly from those taken of CMN patients, especially for young children.

Biography

Ms. Michelle Lee is a student at Thomas Jefferson High School for Science and Technology in Alexandria, Virginia, USA. In the summer of 2011, she had an internship at Zhongshan Ophthalmology Center of Sun Yat-sen University where she shadowed Dr. Guo Xiang-ming on both clinical patient office visits and ophthalmology research. Under the guidance of Dr. Guo, she helped sort hundreds of eye photos taken by the digital-photoscreener, distinguished patients with OA1 disease and CMN disease from normal patients based on the characteristics of the eye photos, and assisted drafting this research paper under Dr. Guo's direction.