

6th Global Ophthalmologists Annual Meeting

May 16-18, 2016 Osaka, Japan

Mutation survey and genotype-phenotype analysis of COL2A1 and COL11A1 genes in 18 Chinese patients with Stickler Syndrome

Xun Wang, Jie Li, Shiqiang Li, Yadi Li, Yantao Wei and Xiangming Guo
Sun Yat-Sen University, China

Purpose: To conduct a survey spectrum of COL2A1 and COL11A1 genes and reveal the genotype-phenotype correlation in Chinese patients with Stickler syndrome.

Methods: A total of 18 Chinese probands with Stickler syndrome were recruited, including nine with a family history of an autosomal dominant pattern and nine sporadic cases. All of the patients underwent full ocular and systemic examinations. Sanger sequencing was used to analyze all of the coding and adjacent regions of the COL2A1 and COL11A1 genes. Multiplex ligation-dependent probe amplification was performed to detect the gross indels of COL2A1 and COL11A1. Bioinformatics analysis was performed to evaluate the pathogenicity of the variants.

Results: Five mutations in COL2A1 were identified in five of the 18 probands, including three novel (c.85C>T, c.3356delG, c.3401delG) mutations and two known mutations (c.1693C>T, c.2710C>T). Of the five mutations, three were truncated mutations and the other two were missense mutations. Putative pathogenic mutations of the COL11A1 gene were absent in this cohort of patients. Gross indels were not found in COL2A1 or COL11A1 in any of the probands. The genotype-phenotype correlation analysis showed that Chinese patients with a family history had more serious ocular phenotypes than the sporadic cases. High myopia was the most frequent initial ocular phenotype of Stickler syndrome. In this study, fourteen Chinese probands lacked obvious systemic phenotypes.

Conclusions: In this study, three novel and two known mutations in the COL2A1 gene were identified in five of 18 Chinese Stickler patients. The results expand the mutation spectrum of the COL2A1 gene. Analysis of the genotype-phenotype correlation showed that patients with a family history had more serious visual prognoses than those of the sporadic cases. Early-onset high myopia with vitreous abnormalities may be a key indicator of Stickler syndrome. Chinese patients had milder systemic symptoms than Caucasians with the same mutation.

zocguoxm@aliyun.com

Minimally invasive glaucoma surgery – Tips on mastering the istent

Robert Chang
Stanford University School of Medicine, USA

This oral presentation will cover tips and pearls on incorporating the Glaukos IStent, a minimally invasive glaucoma surgery (MIGS), into your ophthalmology practice for the treatment of open angle glaucoma. It will encompass an overview of trabecular meshwork-based ab-interno surgery, discuss the new devices with a focus on IStent, introduce gonioscopic angle surgery as a new skill, provide surgical videos of optimal and suboptimal IStent placement, discuss patient selection, technique, efficacy, post-op management and complications and finally, review the literature on IStent outcomes. The course is presented from a United States perspective and briefly mentions cataract surgery in conjunction with the IStent.

viroptic@gmail.com

Notes: