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Reporting Incidental finding of hypertrophic Cardiomyopathy in a patient with RP

Hanieh Noferesti[1], Pardis Ghazizadeh Tehrani[1], Kiyana Sadat Fatemi[1], Tina Shirzad[1], Ameneh Bandehi Sarhaddi[1], Maryam abiri[1,2*], Sirous Zeinali [3,1*]

[1] Dr. Zeinali's Medical Genetics Laboratory, Kawsar Human Genetics Research Center, Tehran, Iran

[2] Departments of Medical Genetics, School of Medicine, Iran University of Medical Sciences, Tehran, Iran

[3]Molecular Medicine Department, Biotechnology Research Center, Pasteur Institute of Iran, Tehran, Iran

Email address: Sirouszeinali@yahoo.com

Mary_abiri86@yahoo.com

Introduction

Retinitis Pigmentosa (RP) is a heterogonous disorder with various mode of inheritance. RP is an inherited progressive retinal dystrophy which lead to loss of the photoreceptors and retinal pigment epithelium which may lead to blindness. Approximately 50% of RP patients don't have a family history of the disease. Herein, we report an extended family suspicious to RP. The causative mutation was found by whole exome sequencing (WES).

Results

WES revealed a heterozygous variant on X chromosome in the RP2 gene, c.534del A (p. Pro179Leufs*59). An additional incidental finding was a heterozygous mutation found in LMNA gene on X chromosome c.1583C>T p.(Thr528Met). All affected members of the family showed the heterozygous mutation of the RP2 gene, healthy members of the family did not show the variant.

Discussion

Next generation sequencing has been made an opportunity to find the causative mutations especially in heterogeneous disorder. But reporting incidental findings is a debate and may complicate the genetic counseling. Both identified variants are inherited in dominant form the mutation in LMNA gene will cause hypertrophic cardiomyopathy which may lead to sudden death. Is it ethical to disclosure of the family the presence of the LMNA gene variant?

Material & method

The proband was a 34 year old woman whose parents are homeland. She was referred to Dr. Zeinali's medical Genetic Laboratory for confirmation of the clinical diagnosis of the RP disease and mutation detection. WES was performed to find the causative mutation. Segregation analysis was performed by Sanger sequencing for other affected and healthy family members.