MATHEWS

Mini-Review

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Plakophilin-2 is Likely to be the Result of Whole Sequencing in Uhl Anomaly

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ABSTRACT

The origin of Uhl anomaly is still unknown. **Method:** In a patient with Uhl anomaly, recently described, genetic whole sequencing was performed. **Results:** At genetic whole sequencing a nonsense heterozygous mutation of plakophilin-2 was identified. Conclusions: This paper offers the first and only positive genetic finding. It is likely that Uhl anomaly is the maximum variant of arrhythmogenic right ventricular cardiomyopathy as plakophilin-2 is the most common mutation in arrhythmogenic right ventricular cardiomyopathy.

Keywords: Uhl Anomaly, Arrhythmogenic Right Ventricular Cardiomyopathy, Plakophilin-2.

INTRODUCTION

Uhl anomaly is extremely rare heart disease with a rate of 1: more than 1000000 inhabitants. There is no information in genetics. We recently reported on a 18-year old male so far asymptomatic patient with all signs of Uhl anomaly [1]. In cardiac MRI the wall of right ventricle was paper-thin and extremely dilated.

METHOD

In this patient genetic whole sequencing was done in the genetic laboratory of the Ruhr Universtiy of Bochum, Germany (Leader: Prof. Dr.med. Hui Phuc Nguyen).

All relevant desmosomal (Plakophilin-2, desmoglein-2, desmocollin-2, desmoplakin, plakoglobin) and non-desmosomal (TMEM43, Lamin A/C, Filamin C, phospholamban) were examined.

RESULTS

In the exom analysis a nonsense variant of plakophilin-2 c.235>T, p.(Arg79#), rs 121434420 was found. This mutation was heterozygous and highly pathogenic (class 5).

DISCUSSION

So far, the cause of Uhl anomaly is unknown. According to these data Uhl anomaly is likely to be the maximum variant of arrhythmogenic right ventricular cardiomyopathy with thinning and dilatation of the right ventricle. This paper offers the first and only positive genetic mutation, a nonsense plakophilin-2 was found. Mutations in plakophilin-2 are the

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main cause of genetically determined arrhythmogenic right ventricular cardiomyopathy wit a rate of 20 up to 40% [2]. Together with the formation of the right ventricle, development of histology and ECG findings [3]. Uhl anomaly is most likely to be the maximum variant of arrhythmogenic right ventricular cardiomyopathy. Of utmost importance is, that arrhythmogenic right ventricular cardiomyopathy can be differentiated from Uhl anomaly with all differences and certain similarities. Plakophilin-2 mutation leads the extreme thinning and dilatation of the right ventricle with missing musculature.

CONCLUSIONS

The finding of the study, that plakophilin-2 is the first and only genetic positive result leads to the theory that Uhl anomaly is the maximum variant of arrhythmogenic right ventricular of biventricular cardiomyopathy.

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None.

CONFLICTS OF INTEREST

The author declares that no conflicts of interest.

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