Medical Expertise

"Development of the European Network in Orphan Cardiovascular Diseases"
„Rozszerzenie Europejskiej Sieci Współpracy ds Sierocych Chorób Kardiologicznych"

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CASE SUMMARY

23-years old, asymptomatic patient with no previous medical history was admitted to hospital to verify suspicion of the Brugada syndrome (BS) diagnosis. Father of the patient had an implantable cardioverter-defibrillator (ICD) implanted as a secondary prevention of sudden cardiac death. He has had a BS type I diagnosed after undergoing ventricular fibrillation with successful reanimation. The patient was diagnosed with Ist degree atrio-ventricular (A-V) block. Holter-ECG revealed 29 pauses, with longest R-R interval of 2387ms, occurring at night and during day activity, in mechanisms of blocked atrial activity, IInd degree A-V block Mobitz I and II. Episodes of isorhythmic atrio-ventricular dissociation with escape nodal rhythm were also registered. EP study revealed prolonged HV distance and low Wenckebach point of 80/min. Visual studies showed no structural abnormalities of the heart muscle.

In conclusion, BS was excluded, however PCCD (progressive cardiac conduction disease) suspicion was raised. Both diseases have common genetic background.

DISCUSSION

Progressive cardiac conduction disease (PCCD) is diagnosed in presence of unexplained conduction disturbances in the conduction system of young (<50 years old) patients without structural heart or muscle diseases. In Brugada syndrome tachyarrhythmia (VT) provocation in EP study has a prognostic value in asymptomatic patients (risk stratification). Family history of sudden

cardiac death and SCN5A mutations have no prognostic value in risk stratification. Pacemaker implantation in PCCD is recommended in intermittent or permanent advanced and IIIrd degree A-V block, symptomatic IInd degree A-V block Mobitz I and II.

ICD implantation in PCCD is recommended in presence of A/C mutation of lamine gene with heart muscle dysfunction, and/or non-sustained ventricular tachycardia.

EXPERT'S OPINION

In young healthy people, with recreational level of physical activity, A-V conduction disturbances are not rare and most commonly are associated with vagotomy. A-V block occurs almost always at night. In the described case distal A-V conduction aberrations were present. PCCD suspicion may be supported by the patient's young age, lack of structural heart disease or myopathy, A-V conduction disturbances, father's diagnosis of BS. SCN5A mutations are responsible for most of the cases of familial PCCD and frequently BS phenotype is present. Maybe the patient and his father share the same SCN5A loss of function mutation but with different expressions.

CONCLUSION

This young asymptomatic patient requires recurrent Holter ECG examination to assess progression of conduction disturbances and detailed genetic examinations (SCN5A, TRPM4, LMNA). At the moment he does not require pacemaker or ICD implantation.

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