BRUGADA SYNDROME: CLINICAL FEATURES, DIAGNOSIS, AND MODERN APPROACHES TO TREATMENT

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Brugada syndrome (BS) is a rare inherited disorder characterized by abnormal electrical activity in the heart, particularly manifested as ST-segment elevation in the right precordial leads of an electrocardiogram. This condition follows an autosomal dominant inheritance pattern and results from mutations in the SCN5A and GPD1-L genes. These mutations lead to dysfunction in Na-channels in the right ventricular cardiomyocytes, causing defective repolarization and arrhythmias. BS is a common cause of sudden cardiac death in individuals without structural heart abnormalities, especially in young adults aged 30 to 45 years [1][3].

Despite the rarity of sudden cardiac death (SCD), this syndrome is a significant factor in sudden cardiac death, which most often occurs during sleep, when the activity of the parasympathetic nervous system predominates. Symptoms of the disease can vary, but most often they are sudden loss of consciousness or even MS, which can be prevented only with early diagnosis. The diagnostic criteria for Brugada syndrome (BS) involve distinct ECG findings, such as a prolonged PR interval, a right bundle branch block pseudoblock, and ST-segment elevation in leads V1 to V3 [2][4]. The ST-segment elevation can appear in various forms, including saddle-back and coved patterns, which are crucial for confirming the diagnosis [4].

Several factors can trigger the onset of Brugada syndrome (BS) symptoms, including hyperthermia, alcohol intake, and the use of certain medications like flecainide, propafenone, propranolol, verapamil, among others. To diagnose the condition, pharmacological testing with novocainamide or flecainide can help identify latent forms of the disease [3][4]. During these tests, characteristic ECG changes are frequently observed, such as dome-shaped ST elevation or widening of the QRS complex.

There are also certain clinical forms of the disease. The most common is the classical form of BS, which is manifested by hyperthermia, as well as other symptoms such as bilateral conjunctivitis, changes on the lips or in the mouth, erythema of the palms or feet, and polymorphic exanthema on the trunk. In rare cases, an atypical form of the disease may occur, where these symptoms may be less severe [1][2].

Managing Brugada syndrome involves a multifaceted approach. Currently, the only established method for preventing life-threatening arrhythmias is the implantation of an implantable cardioverter defibrilator. Pharmacological treatment includes the use of drugs such as quinidine and phospodiesterase III inhibitors, but further research is needed to confirm their effectiveness in treating patients with BS. In some cases, radiofrequency ablation of ventricular ectopy is also used [3]. In addition, an important part of treatment is continuous ECG monitoring and additional tests to assess the risk of arrhythmias.

Brugada syndrome is clinically important because it can cause sudden cardiac death, especially in patients who do not have overt signs of structural heart disease. Therefore, early detection, monitoring and timely treatment are key to reducing the risk of fatal outcomes. Prospects for further research focus on improving diagnostic methods, including genetic tests, as well as developing new therapeutic approaches aimed at correcting the underlying mechanisms of the disease [2][3].

Sources

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