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

The young male was diagnosed with sarcoidosis three ears ago and subsequently one year ago with cardiac sarcoidosis. After initiation of corticosteroids therapy significant improvement in symptoms and exercise capacity was observed. The results of both, baseline ECG and 24-tape were normal. Similarly, objective assessment of exercise capacity, revealed excellent physical condition. Moreover, his echocardiogram was fairly good with normal cardiac chambers size, preserved ventricular function, and no valvular pathologies. However, there were typical changes of gadolinium enhanced pattern in cardiac magnetic resonance.

DISCUSSION

Cardiac sarcoidosis (CS) is an infiltrative, granulomatous disease of the myocardium. Post mortem studies indicate that cardiac involvement ranges from 20 to 25%. At early stages of the diseases majority of CS is clinically silent. Cardiac sarcoidosis drastically changes functional and vital prognosis.

Cardiac involvement affects ≤40% of patients with sarcoidosis and accounts for ≤25% of deaths. It affects cardiac mechanics, causing ventricular failure, and disrupts the cardiac electrical system leading to third degree heart block, malignant ventricular arrythmias, and sudden cardiac death.

The diagnosis of CS is difficult to make. There are clear indications for screening of CS in patients with extra-cardiac disease. In the beginning of diagnostic process the usual steps of detailed history, physical examination, ECG and chest X-ray play a role. The next step is a detailed echocardiography with assessment of systolic and diastolic function, wall motion abnormalities and ventricular wall thickness. Holter monitoring is of paramount importance. More than 100 ventricular ectopic beats during 24-hour tape is a valuable screening criteria. The most advanced imaging and functional modalities include cardiac magnetic resonance imaging (cMRI) and cardiac 18-fluorodeoxuyglucose positron emission tomography (FDG-cPET). Early enhancement of sarcoid granulomas in T2-weighted gadolinium images suggests the presence of inflammation and edema, whereas late enhancement indicates fibrotic changes and scaring. The typical location of morphologic

changes is mid-myocardium of the basal segment of inter-ventricualar septum and lateral wall. In CS cardiac FDG uptake is increased even when other changes are absent in cMRI or other imaging tests. Endomyocardial biopsy (EMB) is the most specific method for the detection of CS. The obvious limitation is sample error that decreases the diagnostic yield of the test. To overcome this issue, the image-guided biopsy is a new, promising tool.

Although there are no published clinical consensus guidelines or systematic evaluation supporting the use of corticosteroids for the treatment of cardiac sarcoidosis, the detection of active CS is commonly accepted state that requires corticosteroids. In the daily practice, the common indications for prolonged corticosteroids treatment are left ventricular ejection fraction < 50%, advanced heart block, ventricular arrythmias, and positive cardiac biopsy. In one study it has been reported that in 57 patients with AV conduction disease who were treated with corticosteroids, almost half of the significantly improved (27/57). Patients with CS are at high risk for ventricular arrhythmias. Implantable cardiac defibrillator (ICD) implantation is a class IIA recommendation for patients with CS. On the other hand this population also has high rates of inappropriate shocks and device complications. Young patients with progressive cardiomyopathy should be considered for the evaluation for heart transplantation.

EXPERT'S OPINION

The presence of cardiac sarcoidosis in the presented patient is questionable. The only abnormality detected is abnormal gadolinium enhanced pattern in cardiac magnetic resonance, whereas all other imaging and functional tests are normal. The significance of pure CMR changes in patients with sarcoidosis is unknown but probably represents benign situation. Unfortunately, data on pulmonary and other organs involvement have not been provided. On the other hand, the patient has been treated with corticosteroids for a long time. From the cardiac perspective, there are clear indications for corticosteroids or other cardiac-specific agents. Nevertheless, the patient should be periodically monitored with preferentially ECG, Holter and echocardiography. It is difficult to provide the specific time frame but to be on the safe side ECG should be performed during each office visit, Holter every six months and echocardiograpy once a year.

CONCLUSION

Cardiac sarcoidosis is an uncommon diseases. Nevertheless, when present dramatically increases mortality. Early diagnosis and treatment can be beneficial and is cost-effective. The presented patient is asymptomatic from cardiac perspective. Moreover, his imaging and functional cardiac tests are normal, except from some abnormalities detected in CMR. Consequently, there are no indications for the prolonged cortisteroids to treat cardiac condition. On the other hand there may be some other extra-cardiac indications, that should be appropriately followed. At present the patient required only periodical cardiac follow-ups.

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