Material and Methods During a two year interval, a number of 6 adolescent males were admitted in the ED with severe precordial pain. All were between

14-17 yo. They performed lab investigations, ECG, Echocardiography, Speckle-tracking, Holter ECG and cardiac biomarkers. Two were explored by angiocoronarography due to acute STEMI aspect on ECG, to exclude myocardial infarction. Angio MRI of the heart performed in all patients.

Results Six males were admitted for severe precordial pain, 8 on a scale of 0-10, which appeared within the last 12 hours, with a duration of 1-2 hours, accompanied by palpitations. One boy had Duchenne disease and one thrombophilia. The CRP was slightly elevated in all cases, with values between 7 and 56 mg/dl; hsCRP was also elevated. CPK was increased in all cases, varying between 134 – 1834 ng/ml. The highest value was in Duchene patient. CPK-MB was slightly increased in 4 patients and very increased in two. Troponin T was positive in two patients with values 54 and 51 pg/ml and increased values in four, between 669 and 2500 pg/ml. Troponin I had very high values, from 501 to 30.000 pg/ml in one case of multifocal myocarditis.

NTproBNP was altered in all patients, with values between 72 and 1508 pg/ml.

TGO and TGP were elevated, especially TGO. ECG in all cases had the aspect of acute STEMI. Two cases were explored by angiocoronarography with negative result. Ejection fraction was at the lower limit of normal in 3 cases and reduced between 45-49% in the other three. Speckle-tracking was significant in all 6 cases. Holter ECG detected PVC in 2 cases and bradycardia in one.

Cardiac MRI revealed inflamed heart muscle at late gadolinium enhancement.

MRI was repeated at 6 mo and 1 year after, with good results. Viral serology was negative for: parvovirus, coxackie, adenovirus, cytomegalovirus.

Treatment included: AINS, Carvedilol, diuretic, vitamins, bed rest until troponin normalization and sport avoidance 6 mo to 1 year.

Conclusions Myocarditis in adolescents almost always mimics myocardial infarction. Cardiac biomarkers help in the diagnosis, evolution and follow-up. Troponin I is more accurate than troponin T. The extremely high values of Troponin I was correlated with severe acute multifocal myocarditis. Cardiac MRI with late gadolinium enhancement is the hallmark of myocarditis.

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ARRHYTHMOGENIC CARDIOMYOPATHY IN A 15-YEAR OLD BOY WITH DIZZINESS AND SYNCOPE

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10.1136/archdischild-2021-europaediatrics.174

Arrhythmogenic cardiomyopathy (arrhythmogenic right ventricular dysplasia) is a hereditary heart muscle disease characterized pathologically by dystrophy and fibrofatty replacement of the right ventricular myocardium, consequently causing ventricle dysfunction and life-threatening cardiac arrhythmias. The right ventricle is predominately affected, but approximately 70% of hearts studied during autopsy reveal left ventricle involvement. Positive family history is seen in 30% cases.

Autosomal dominant or autosomal recessive pattern of inheritance are known, latter as part of cardiocutaneous syndromes. The mean age of diagnosis is 30 years, clinical manifestation is rare before puberty. Following hypertrophic cardiomyopathy, it is considered to be the leading cause of sudden cardiac death in adolescence, especially among athletes.

Palpitations, dizziness and syncope can be symptoms of the disease, though sudden cardiac arrest may be the first clinical presentation. Diagnosis is made by combining medical history, ECG, morphological abnormalities (echocardiography and/or cardiac MRI), endomyocardial biopsy and genetic tests. Standardized diagnostic criteria (major and minor), revised in 2010 were made to increase sensitivity and specificity. Treatment options are combination of life-style modification, antiarrhythmic drugs, catheter ablation and ICD implantation.

Here we are illustrating a 15-year old boy who presented with dizziness and syncope. Cardiac diagnostic workup was performed, revealing arrhythmogenic cardiomyopathy. Inverted T waves in V1-V4 leads with epsilon waves and premature ventricular contractions with LBBB were noted in standard ECG.

During the 24-hours holter ECG sustained ventricular tachycardia was registered. Echocardiography and cardiac MRI revealed hypertrabeculated right ventricle with regional dyskinesia and akinesia, dyssynchronous right ventricle contractions and additionally fibrous replacement of the right myocardial free wall. ICD was implanted after inadequate antiarrhythmic drugs response.

Conclusion cardiac diagnostic workup should be performed as a severe and progressive heart muscle disease can be the underlying cause of dizziness and syncope.

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VERY RARE CASE OF NOONAN SYNDROME, TYPE 2

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10.1136/archdischild-2021-europaediatrics.175

Background Noonan syndrome, type 2 (NS2) is rare autosomal recessive disorder of RASopathies group, caused by mutations in the LZTR1 gene. NS2 characterized by a typical face, short stature, broad, short neck, congenital heart disease, developmental delay. The most common heart disease in children with NS2 is hypertrophic cardiomyopathy.

Patients and Methods Patient is a boy, 15 years old with short stature, developmental delay at 1 first year of life and heart disease. He had distinctive facial features of NS: downsloping palpebral fissures, epicanthic folds, hypertelorism, low-set ears short neck, wing-like folds on the neck, pectus deformity. Hypertrophic cardiomyopathy was identified at 1 month old. Now patient have obstructive, hypertrophic cardiomyopathy, cardiac arrhythmia: ventricular extrasystoles, 4A Lown, intraventricular block combined with bundle branch block, transient WPW. Syncopal episodes.

Surgical correction of obstructive hypertrophic cardiomyopathy: septal myectomy was performed on the child due to the high risk of developing sudden death syndrome. Target areas of the exome were investigated by next generation sequencing (NGS). Bioinformatic analysis was carried out using ACMG recommendation. Validation of the identified variants was carried out by the Sanger method.

Results We revealed nucleotide missens VUS: c.1259A>G, p.Q420R and c.2051T>C, p.I684T in the heterozygous state in LZTR1 gene. All variants were absent in HGMD professional and genome aggregatioonal database.

Conclusion Child with severe hypertrophic cardiomyopathy and typical phenotype of Noonan syndrome was detected NS 2, caused by compound heterozygous missense variants c.2051T>C, p.I684T and c.1259A>G, p.Q420R in LZTR1 gene.

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ECHOCARDIOGRAPHIC FINDINGS IN TUBEROUS SCLEROSIS COMPLEX-SINGLE INSTITUTION EXPERIENCE

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10.1136/archdischild-2021-europaediatrics.176

Tuberous sclerosis complex (TSC) is a relatively rare autosomal dominant disorder characterized by hamartomatous lesions in a variety of organs.

Cardiac involvement remains within major criteria for diagnosing the syndrom, as it can initiate the TSC evaluation for some patients. Cardiac involvement is usually maximal at birth or early infancy and shows signs of regression during the first two years of life. The aim of this study was to evaluate the echocardiographic findings in our patients with tuberous sclerosis complex.

We present a case series of five patients diagnosed with TSC in our hospital within the last ten years. All patients underwent a complete cardiac examination at the time of diagnosis (clinical findings, electrocardiography and ultrasound examination of the heart) and were further monitored through controls.

All patients had a normal clinical findings. One patient had nonsignificant supraventricular and ventricular extrasystoles. Abnormal echocardiographic findings were seen in four of total five patients. Two patients had both atrial and ventricular cardiac masses with echo characteristics that raised suspicion of rhabdomyoma. Two other patients had multiple focal areas of increased intramyocardial echogenicity. There were no signs of outflow tract obstruction or impaired valvular function. Left ventricular size and function were normal. Close follow up showed no signs of significant regression or progression in our echocardiographic findings.

The prognosis and growth potential of these masses are not known, but can be determined by longitudinal follow-up. Cardiac ultrasound should be considered for all patients with TSC regardless of physical findings.

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RARE CASE OF UHL'S ANOMALY IN AN ADOLESCENT

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10.1136/archdischild-2021-europaediatrics.177

Uhl's anomaly is an extremely rare congenital heart defect of an unknown cause, characterized by the absence of the myocardium in the right ventricular free wall, which is replaced by nonfunctional fibroblastic tissue.

We present a case of 14 year old boy after heart transplantation and care due to Uhl's anomaly diagnosed five years ago. Five years before this admission, child experienced exertional dyspnea and vertigo at school.

During transport to the hospital, child arrested and after a short resuscitation and defibrillation (initial rhythm VF), sinus rhythm occurred.

On the initial transthoracic echocardiography, dilated cardiomyopathy of right ventricle was observed. After the initial stabilization, MR was performed (May 2014.), where marked dilatation and impaired function of both right atrium and ventricle was noticed. The volume of right ventricle in diastole (EDV) was 245 ml (188ml/m2) with ejection fraction of RV of 19%.

The RV wall was thin, with almost complete absence of right ventricular free wall myocardium, without fat infiltration. Control MR, performed after one year, showed a progression in dilatation of right ventricle dimensions with impressive tricuspid regurgitation. The left heart structures, functionally, and morphologically, were within normal range. Differential diagnosis yielded two possibilities: Uhl's anomaly and arrhythmogenic dysplasia of right ventricle (ARVD). The recommended biopsy, as well as installation of implantable cardioverter defibrillator, were rejected by his parents. The metabolic diseases were excluded. From June 2018, child was dyspneic in everyday activities, had peripheral edema, was tired and sometimes complaining off tingling in the legs. After two rejections from his parents, heart transplantation was done in January 2019. Pathohistological diagnosis of Uhl's anomaly was confirmed. One month later, he was discharged from the hospital.

It is a question of differentiation between Uhl's anomaly and ARVD. In ARVD, the myocardium is progressively replaced by fibrofatty tissue, and has been linked to mutations in the genes encoding plakoglobin and desmoplakin.

Patients with Uhl's anomaly rarely survive to adulthood. There are two main surgical approaches for these patients; first one is based on the relief of right ventricle volume or excluding the entire right heart from circulation. This can be done by making total cavopulmonary connection, by closing tricuspid valve and making bidirectional Glenn procedure, or making a one-and-a-half ventricle repair with plication of the right ventricular free wall. Second option is the heart transplantation.

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PERIOPERATIVE MYOCARDIAL PERFORMANCE IN INFANTS WITH DOWN SYNDROME UNDERGOING CONGENITAL HEART DISEASE REPAIR: A COMPARISON WITH INFANTS WITH A NORMAL CHROMOSOME COMPLEMENT

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10.1136/archdischild-2021-europaediatrics.178

Infants with Down Syndrome (DS) undergoing surgical correction of congenital heart disease (CHD) may exhibit altered loading conditions and myocardial performance during the peri-operative period when compared with counterparts with a normal chromosome complement. This may result in higher morbidity. We aimed to characterise the impact of DS on myocardial performance and loading conditions in infants with