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Case Report

From shadows to spotlight: how two cases redefined Barth syndrome awareness in Belarus: two case reports with a literature review

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ABSTRACT

Barth syndrome (BTHS) is an uncommon congenital sex-linked recessive cardiovascular disease resulting from impaired cardiolipin metabolism. The main cause of this condition is a mutation of the TAZ gene (TAFAZZIN), which activates the mitochondrial enzyme acyltransferase/transacylase, necessary for the biosynthesis of cardiolipin. The frequency of BTHS is 1 case per million men, and the total number of reported cases worldwide is about 250. The article presents 2 cases of BTHS diagnosis in the same city. A 9-year-old boy who was admitted to the intensive care unit of the Grodno Regional Children's Clinical Hospital was identified in pediatric observation. In the case of an adult patient, an 18-year-old boy was hospitalized in the Grodno Regional Cardiology Center with an exacerbation of heart failure. In both cases, cardiovascular complications were identified when an adult had an atypical phenotype of cardiomyopathy-dilated right ventricular cardiomyopathy and a broad spectrum of comorbidities, and a pediatric patient had visible cardiomegaly, which may be associated with changes in energy metabolism in the heart over time, as evidenced by imaging results. In addition, both patients underwent thorough genetic testing, which confirmed the diagnosis of BTHS in these two cases. This article illustrates the importance of raising awareness about BTHS in the Eastern European region.

Keywords: Barth syndrome, TAZ gene mutation, Cardiomyopathy cardiolipin metabolism

INTRODUCTION

Barth syndrome (BTHS) is ultra-rare congenital X-linked, recessive cardiovascular condition originated from a mutation of TAZ (TAFAZZIN) gene, which is important in encoding the mitochondrial enzyme acyltransferase/transacylase required for biosynthesis of cardiolipin. This disease was first depicted by Barth et al in a large descent of Dutch patients.[1] This syndrome mainly comprises of cardiomyopathy, musculoskeletal myopathy, growth restriction, neutropenia and 3-methylglutaconic aciduria type II.[2] The frequency of occurrence of BTHS is one in millions of males with case totality of approximately 250 worldwide.[3] The role of cardiolipin is diverse in the cell mitochondria consisting of activity of respiratory chains, stabilization of respiratory chain supercomplexes, cell apoptosis regulation and morphology of cristae of mitochondria, thereby maintaining the dynamic system of mitochondria.[4-7] The manifestation of organ-specific symptoms is currently

unclear, but a molecular insight into tissue-specific cardiolipin acyl chain population is depicted, where composition of cardiolipin specific tissues such as heart (80-90% linoleic acid moieties), brain (consist of a wider range of acyl chains) and blood.[8] The epidemiological demography of BTHS is relatively homogenous, with known cases originating from every continent (America, Europe, Japan, Australia) globally. In the perspective of gender, BTHS is predominantly presented in males than females, due inactivation of recessive trait by the other X-chromosome in females, thus in most cases, females were carriers of this syndrome (X-linked recessive pattern.[9] Furthermore, all the reported cases of BTHS patients were males, until 2012, where a female case of BTHS was presented with this critical heart failure at 1 month of age, even though this occurrence was novelty in theory. The clinical presentation of this female BTHS is similar in comparison to usually affected males. Furthermore, clinical illustration of females necessitates the hemizygosity of already mutated alleles of TAZ gene, where a significant rearrangement of alleles is present in the other X-chromosome.[10] The BTHS affects the following systems such as cardiovascular system with clinical manifestations like: congestive cardiomyopathy, noncompaction cardiomyopathy, hypertrophic cardiomyopathy, mixed cardiomyopathy phenotype, variable form of cardiomyopathy, endocardial sclerosis, ventricular arrythmias leading to sudden cardiac death; metabolic and endocrine system with the following clinical findings: 3-MGCA aciduria, delayed skeletal maturation, stunted growth, constitutional delayed puberty, osteopenia, hypocholesterolemia; Hematological System with the following clinical manifestation of neutropenia. Other systems affected were musculoskeletal system with clinical features such as proximal myopathy, easy fatigability, exercise intolerance and maternal-fetal abnormalities such as miscarriage/stillbirths, cardiomyopathy, heart failure/hydrops were recorded as well.[11]

In terms of diagnosis of BTHS in patients, the following methods consisting of clinical presentation with the major clinical features such as suspected male, cardiomyopathy, neutropenia, skeletal myopathy and elevated levels of urine 3-methylglutaconic acid. Furthermore, instrumental diagnostic techniques such as echocardiogram, cardiac MRI were helpful in revealing any early signs of cardiomyopathy (either dilated, hypertrophic or mixed phenotype) in BTHS patients.[11] Another important diagnostic assay became the pioneer in early diagnosis of BTHS in patients, which calculates the MLCL/L4-CL ratio (monolysocardiolipin/tetralinoleoyl-cardiolipin) in blood of BTHS patients. Currently, this is the gold standard for diagnosis of BTHS.[12] In addition, the molecular genetic testing was implemented to identify the hemizygous pathogenic variant in TAZ gene. And the diagnosis of female patients with BTHS, the following combination of suggestive clinical findings and molecular genetic testing of pathogenic variant of TAZ gene is implemented.[13]

The management of BTHS is a multidisciplinary approach with key points to taken into account such as treatment of clinical manifestations, prevention of secondary clinical complications, 24-hour supervision, agents to avoid, evaluation of risk factors in relatives and management in pregnancy.

This article illustrates the importance of how two cases of BTHS recorded in Grodno region of Belarus improved the awareness of BTHS in eastern Europe, thus improving the standard management against BTHS and implementation of earlier interventions for a better prognosis in patients with BTHS.

CASE REPORT

Case 1 (pediatric case)

The patient, a 9-year-old boy, was admitted to the emergency treatment unit to the Regional Clinical Pediatric Hospital in Grodno with acute onset lethargy, pallor, facial puffiness, and labored respirations. The symptoms started one day prior to admission with periorbital edema, which was initially diagnosed as angioedema and treated with prednisolone in the ambulance, thus temporarily improving the condition. The prescription of Suprastin administered subsequently for 2 days. However, on the third day of illness, the patient developed signs of severe dyspnea and oliguria, thereby, necessitating the emergent hospitalization.

The past medical history of this patient revealed term birth (birth weight 2850 g, length 50 cm), but developmental milestones were noticeably delayed. In addition, the growth of patient was in the suboptimal range, with a recorded weight of 7.2 kg at 1 year and 10 kg at 3 years. Furthermore, parents reported that the patient had a history of exertional fatigue and reduced exercise tolerance from an early age.

Significant past medical events included chronic fluctuating neutropenia (absolute neutrophil count ranging from 0.5×10^9/l to 0.8×10^9/l) documented over a three-year outpatient observation period, which tend to normalize during intercurrent infections. At 6 months of age, he required inpatient treatment and surgical drainage for a carbuncle of the upper lip. During his first year of life, he experienced three hospitalizations for acute bronchitis, pneumonia, and obstructive bronchitis. Notably, the previous chest radiographs obtained during these admissions had demonstrated signs of cardiomegaly (with a cardiothoracic index of 56%). An immunology consultation in infancy did not identify any primary immunodeficiency. But patient's family history was significant for maternal juvenile idiopathic arthritis, which was diagnosed at the age of 12-14 years. This condition was previously managed with the application of glucocorticoid therapy. Other siblings including the elder sister was reported to be healthy.

Lab findings

The complete blood count did not reveal any significant deviation from the normal values of some blood elements and markers.

Imagine and instrumental findings

The echocardiogram revealed a dilation of the left heart chambers with an EF of 39% and moderate hypertrophy of the left ventricular myocardium, mainly in the posterior wall of the left ventricle. In addition, mitral regurgitation stage 2-3 was noticed against the background of dilation of the left ventricle and the mitral valve ring.

On the chest X-ray, the heart is sharply dilated in the cross-section with a Cardiothoracic Index (CTI) of 65% in Figure 1.

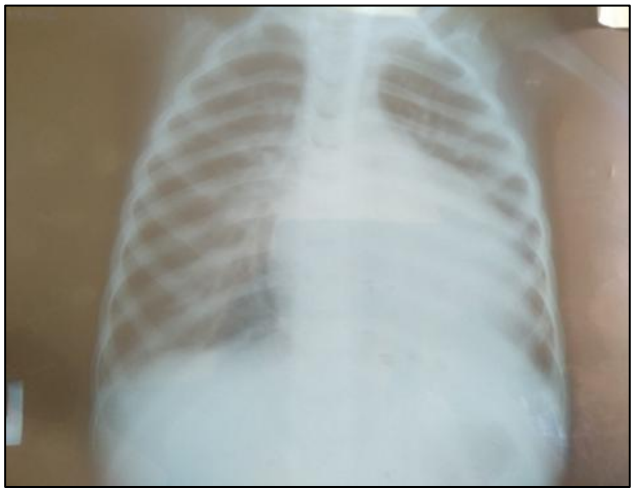

Figure 1: The chest X-ray of patient 1 showed cardiomegaly with a CTI of 65%.

During the hospitalization, signs of heart failure persisted despite pharmacological therapy, the following symptoms were observed such as dyspnea with a frequency of 36-40 per minute, tachycardia of 140-150 beats per minute and periodic gallop rhythm. In addition, there were an increase in the size of the liver (+3.0 cm), spleen (+2.0 cm) compared to the age-related normal values. Furthermore, the diuresis was within 1.6-2.0 ml/kg per hour against the use of a diuretics.

It was recommended to continue administration of digoxin with a dose of 0.01 mg/kg per day, enalapril 0.1 mg/kg per day, furosemide 1 mg/kg per day and aspirin 5 mg/ kg once a day with dose adjustment as the patient's weight increases. In order to clarify the diagnosis, the child was examined in the laboratory of non-chromosomal heredity of the Institute of Genetics and Cytology of the National Academy of Sciences of Belarus. The results were collected by the targeted NGS method using TruSight Cardio Sequencing Kit (Illumina Inc., USA) and verified by Sanger sequencing. The patient had a splicing mutation c.239-1_239delinsTT (NM_000116) in the 3rd exon of the Tafazzin gene.-TAZ (Xq28). These changes of this gene were associated with the development of BTHS. The mutation identified in this patient was not depicted in the clinical genetic databases (NCBI ClinVar, HGMD as of 11.05.2020). In addition, further analysis was carried out as part of research work with the informed consent of the patient and his legal advisors. The following diagnosis of BTHS with X-linked recessive inheritance pattern was confirmed.

Case 2 (adult case)

The patient, an 18-year-old boy presented to the Grodno Regional Cardiology Center with a 3-month history of worsening exertional dyspnea, fatigue, and lower limb edema. Since age 16 (2022), the patient had experienced symptoms and signs of heart failure, including exertional dyspnea, fatigue, chest pain, nosebleeds, and leg cramps and subsequently diagnosed with Arrhythmogenic cardiomyopathy (right dominant variant) and secondary tricuspid valve insufficiency. The patient was also identified with a reduced ejection fraction of the left (34%) and right (17%) ventricles. Cardiac MRI also revealed extensive fibrosis in right ventricle and biventricular dysfunction and noncompact myocardium. About two months before this visit, a Holter monitor recorded 589 premature ventricular beats and two episodes of non-sustained ventricular tachycardia, with a maximum rate of 175 beats per minute, leading to the patient's admission into the intensive care unit.

The developmental history revealed that the patient was born full-term in April 2006 (weighing 3,050 grams) with an Apgar score of 8,9. In addition, there were signs of delayed early motor milestones and the patient received treatment at around two months for motor developmental delay related to neonatal encephalopathy.

The physical growth of patient had been inadequate for his age (height 168 cm, weight 52 kg, BMI 18.4 kg/m²), and presented with a characteristic "Angelic face" (a high, broad forehead and narrow chin). The cognitive development was normal, and the patient is currently studying in a technical college.

The patients' family history depicted that the patient was part of a twin pregnancy; unfortunately, the latter died in utero. The patient's mother has WPW syndrome and treatment was proceeded with radiofrequency ablation. The patient's father had no known heart abnormalities. Other siblings like two sisters presented with celiac disease, but no reported heart problems.

The past medical history of the patient revealed on the second day after birth, ECG depicted a heart repolarization abnormality, and chest X-ray revealed cardiomegaly. The patient was diagnosed with "morpho-functional immaturity". In addition, there was a presence of mild mitral valve insufficiency and abnormal cord anatomy in

left ventricle. Furthermore, at the age of six-month mark, the patient was hospitalized with immune neutropenia, congenital carditis, mild anemia, and frequent infections during childhood.

Despite initiation of medical treatment, patient reported to have frequent infections; which includes acute respiratory infections, streptodermia, tonsillitis, otitis and furuncle. At age of 6, the diagnosis of short stature, celiac disease and subclinical hypothyroidism were clinically proven.

The ECG (Figure 2) of this patient depicted the following results: a sinus rhythm, regular in nature, with a heart rate of 96 bpm, right axis deviation, signs of hypertrophy and overload of the right atrium. Depolarization disorders were detected in (widened QRS in lead V 2, ϕ (epsilon) chest leads V1 -V2, decreased QRS voltage (V3, V4) and repolarization (ST-T changes in most leads).

Lab findings

The complete blood count (CBC) shown in Table 1 did not reveal any significant deviation from the normal values. But there was a noticeable decrement of neutrophil count to 37% suggesting one of the classical signs of BTHS, which is Neutropenia. Furthermore, the level of monocytes (13%) and lymphocytes (48%) were significantly high, suggesting an underlying inflammatory response from the body to an infection.

The biochemical analysis shown in Table 2 revealed elevated liver enzymes (AST and ALT), total bilirubin and lactate dehydrogenase, a suggestive feature of poor cardiolipin synthesis; observed in BTHS, thus rupture of hepatic cells and erythrocytes respectively.

Imaging and instrumental findings

According to the cardiac MRI of this patient (with contrast enhancement by gadolinium) revealed a decrease in the ejection fraction of the left (34%) and right (17%) ventricles, a non-compact myocardium along with anterolateral and inferior wall of the left ventricle at the level of the middle segments, extensive areas of fibrosis in the upper, lower and lower-lateral walls of the myocardium of the right ventricle were also shown (Figure 3).

Table 1: Complete blood count parameters.

CBC parameters	
RBC	5.63 million/ µl
Hb	176 g/l
WBC	4.4 thousand / µl
ESR	2 mm/h
Hematocrit	53%
Platelets	186 *1000/ µl
MCV	94.1 fl
MCH	31.3 pg
MCHC	332 g/l
RDW	14.5%
Colour index	0.94
Eosinophils	1%
Band neutrophils	1%
Segmented neutrophils	37%
Lymphocytes	48%
Monocytes	13%

Table 2: Biochemical parameters.

Biochemical analysis	
Total protein	70.6 g/l
Albumin	36.2 g/l
Urea	6.9 mmol/l
Creatinine	62 µmol /l
Cholesterol	2.3 mmol/l
Total bilirubin	**35.2** µmol /l
Blood glucose	4.8 mmol/l
AST	80.6 U /l
ALT	44.2 U/l
Lactate dehydrogenase	1265 U /l
Sodium	136.4 mmol/l
Potassium	5.2 mmol/l
Chlorides	100 mmol/l

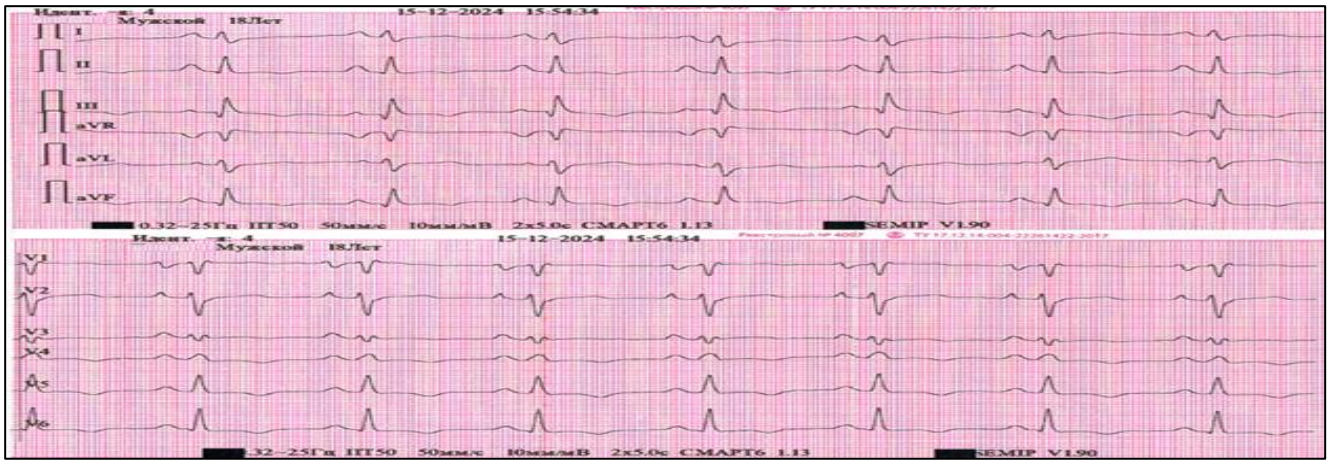

Figure 2: The ECG of this patient 2.

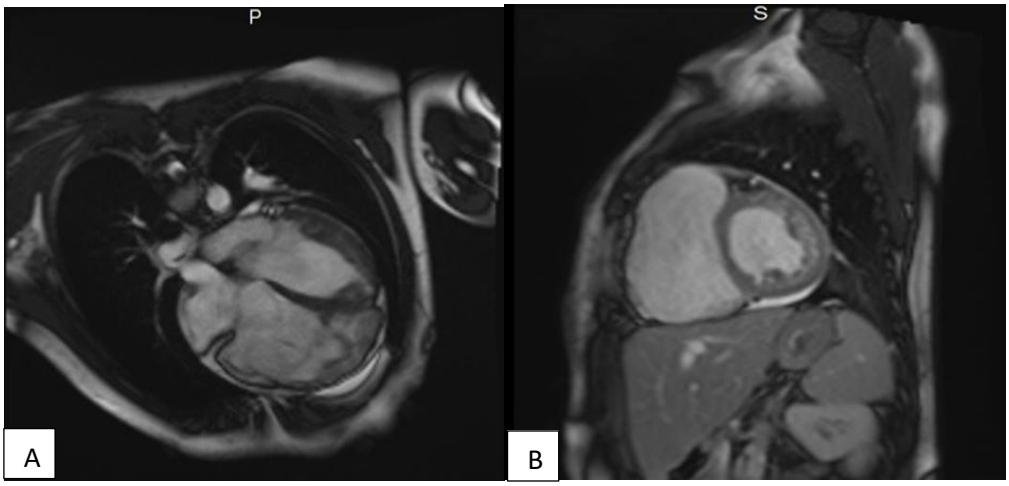

Figure 3 (A and B): MRI of the heart of patient 2 with fibrosis fields were detected in the myocardium. Significant dilation of the right ventricle, right atrium, moderate dilation of the left ventricle was observed.

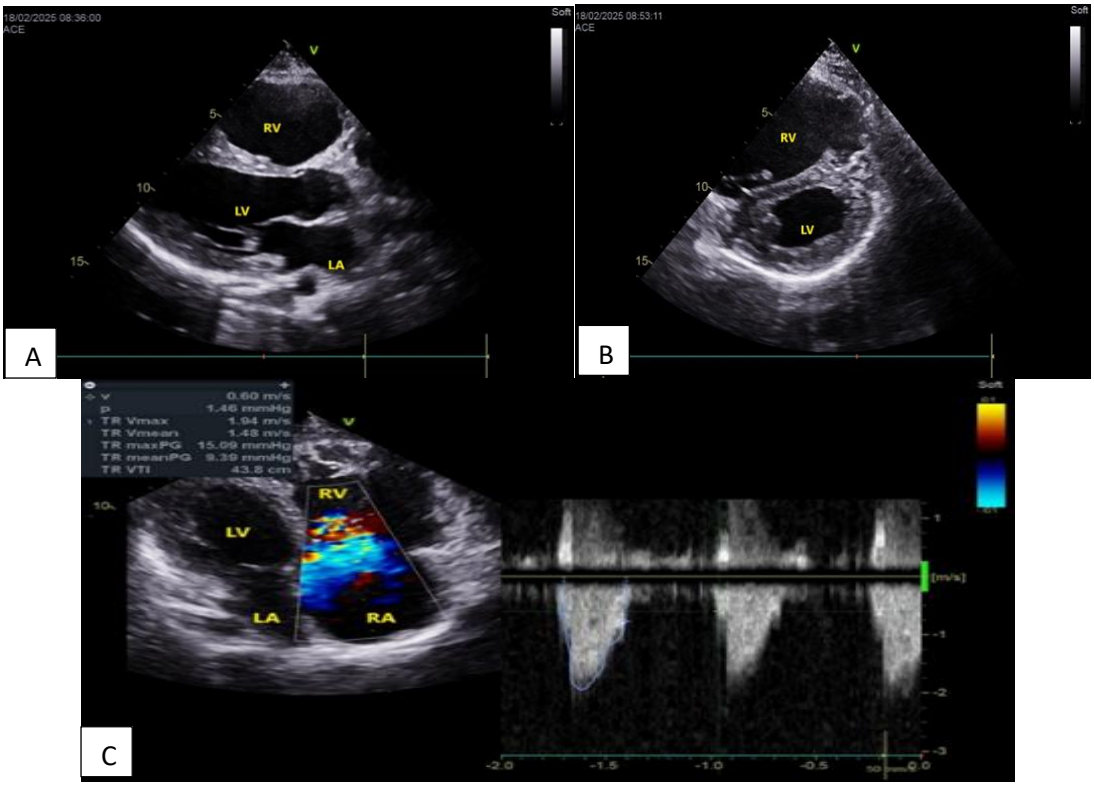

Figure 4: (A-C) The images of echocardiogram of patient 2 in the longitudinal, transverse and 4-chamber view.

The data from echocardiogram revealed pronounced dilation of the right chambers of the heart, increased trabecular pattern of the right ventricle, thinning of its wall, and decreased contractility of the right ventricle.

The Holter monitoring (HM) of the ECG revealed a single episode of tachycardia lasting a period of 10 seconds with a frequency of 175 per minute, without any indication of the origin of the tachycardia.

Based on the collected data, the council of specialists decided to refrain from surgical correction and implemented drug therapy with enalapril, furosemide and sotalol. Prevention of infective endocarditis was indicated and, also probability of life-threatening arrhythmias was high. Thus, the implantation of a cardioverter-defibrillator was recommended. Furthermore, the patient had been subsequently placed on a heart transplant waiting list and was recommended to the undergo genetic testing of the heart.

A genetic study was conducted in the clinical diagnostic laboratory of the Mother and Child Republican Scientific and Practical Center, which determined that patient was a

carrier of a mutated variant of TAZ gene in a hemizygous state; mutations in this gene are primarily associated with the development of BTHS with X-linked recessive inheritance pattern (BTHS OMIM 302060. Genotype C/C p.Gly80Arg). Based on the results, the identified variant of the nucleotide sequence was assessed as pathogenic. The diagnosis was confirmed as BTHS.

After reaching 18 years of age, the patient had been monitored by cardiologists at the State Regional Cardiology Center. During this period from 2022 to the present, the patient had experienced an increase in symptoms of heart failure, in the form of significant shortness of breath and fatigue, and the appearance of swelling of the lower extremities. During the HM of ECG following 2 years, the results such as 589 ventricular extrasystoles and 2 episodes of unstable ventricular tachycardia were recorded (max. number of complexes-9, maximum heart rate-175 per minute).

The echocardiography revealed the following findings such as dilation of the cavities of the right atrium (52×68 mm), right ventricle (36 mm) and dilation of the tricuspid valve annulus. This patient was further diagnosed with tricuspid grade 3 regurgitation with moderate pulmonary hypertension (systolic pressure-45 mm Hg) and grade 1 mitral regurgitation.

Furthermore, left ventricular myocardial hypertrophy was found in the echocardiogram. In addition, there was decreased systolic function of the left ventricular myocardium (LVEF-47%) and right ventricle (TAPSE 13 mm) suggesting type 1 diastolic dysfunction of the left ventricular myocardium. And bilateral minor hydrothorax was also observed.

DISCUSSION

BTHS is a potentially fatal genetic sex-linked disorder that influences males with a relatively high infant mortality rate. We present rare cases of one patient with BTHS surviving till adulthood and another of a child with BTHS. BTHS is a multisystem disorder affecting the cardiovascular, neurological, immune, musculoskeletal and metabolic systems; hence the BTHS registry was created with the BTHS Foundation for the ease of clinically identifying features of BTHS.[2,11] According to the data taken from the BTHS Foundation cardiovascular manifestations account for up to 90% including dilated CMP, left ventricular noncompaction, hypertrophic CMP; as evidenced by both our patients showing cardiovascular effects the adult with an atypical phenotype of cardiomyopathy transforming from a rare hypertrophic CMP to a nondilated CMP, and the pediatric patient with a visible cardiomegaly which can be attributed to the changes of energy metabolism in the heart overtime as evidenced by their echo and imaging findings as discussed above in the imaging and diagnosis section of this article. Neurological manifestations in these patients were not very prominent apart from impairments in behavior owing to the long-term course of illness without significant decline of cognitive function. These patients had histories of recurrent chest infections, tonsillitis, otitis; which could be attributed to the neutropenia caused by the disease which is a feature in about 69% of patients with BTHS.[14] Granulocyte colony stimulating factor (G-CSF) has been predominantly utilized in BTHS with a considerable amount of success. G-CSF is generally used in the period of neutropenia concomitant with relevant preventive antibiotics if clinically indicated. Although in our patients only antibiotics were used probably due to lack of readily available factor. The patients showed early signs of exercise intolerance which could either be attributed to having a cardiogenic cause or due to the musculoskeletal manifestations of BTHS. Their pediatric histories are also significant for delay in growth and reaching developmental milestones observed in about 60% of BTHS patients.[15] Late diagnosis or improper treatment of cardiomyopathy in BTHS may progress to heart failure. Longstanding cardiac dysfunction in the adult patient led to exercise intolerance and reduced EF which improved upon the administration heart failure therapy.

This case is a prime example showing the declining course of cardiomyopathy in the absence of pathogenetic therapy and challenges faced in optimizing treatment. Often than not, myocarditis in the background of heart failure may also present as a cardiac manifestation in these patients as was observed in our pediatric patient and hence enalapril and furosemide were prescribed to tackle the fluid overload and cardiac remodeling.

A confirmatory diagnosis of Barth's syndrome is made by genetic testing in TAZ by sequence analysis as was done and confirmed in our pediatric patient in which changes of this gene are associated with the development of BTHS. Multisystem diseases like BTHS pose a significant threat in effective diagnosis and therapy contributing to its increased mortality.

One of the main novel therapies of management for BTHS is a water-soluble, aromatic-cationic mitochondria-targeting tetrapeptide that freely infiltrates the inner mitochondrial membrane where it connects with cardiolipin, improving membrane stability as evidenced by the TAZPOWER trial.[16] Elampiretide has shown positive benefit in most affected systems by the disease including the skeletal muscle, heart and positive mitochondrial dynamics.[17]

CONCLUSION

Even though BTHS is a rare occurrence, its prevalence has increased over the years making the issue of raising awareness about these conditions among the medical providers and the general population important. Therefore, this article also highlights the awareness of BTHS in Belarus, and how it is in real clinical practice medical intervention implemented for management and improve prognosis of both cases of BTHS. Furthermore, it is of

utmost importance to develop widely accessible, affordable and effective ways of gene modification therapy, providing more effective methods to treat for its many lethal manifestations like heart failure and neutropenia for which research plays a major role.

Recommendations

Further genetic and clinical studies are warranted to explore the clinical manifestations of BTHS to improve the treatment strategies in real clinical practice.

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