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Case Report

The Squeezed Heart-A Case Report

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Abstract

Pompe's disease is an autosomal recessive disorder caused by inherited deficiency of α-1,4-glucosidase (acid maltase), a lysosomal enzyme. Patients usually die in the first year of life from cardio-respiratory failure due to massive left ventricular hypertrophy. We report a case of 3-month-old boy presented with fatal infantile onset Pompe's disease.

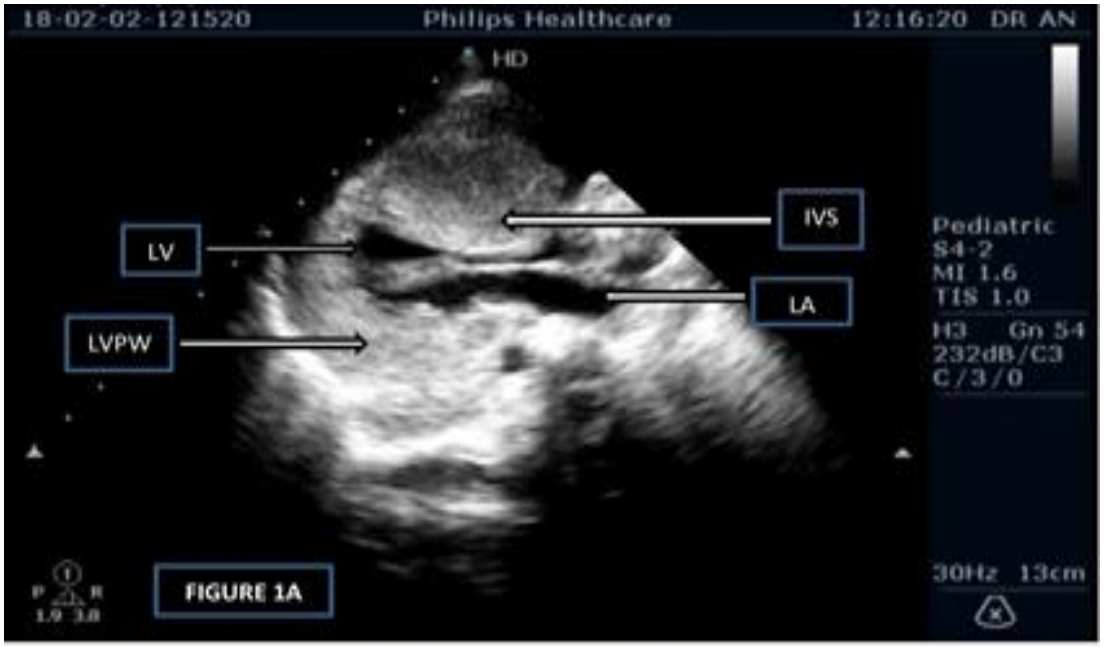
Key Words: pompe's disease; biventricular hypertrophy; glycogen storage disease

Introduction:

Glycogen storage disease type II or Pompe's disease is a rare hereditary error of carbohydrate metabolism in which excessive quantities of glycogen accumulate in the heart, muscle and other tissues [1,2]. Pompe's disease is characterized by infantile onset muscular weakness, hypotonia, and enlargement of the heart and liver, followed by progressive cardiorespiratory failure [2]. We report a case of 3-month-old boy presented with progressive respiratory distress who had infantile onset Pompe's disease.

Case report:

A 3-month-old male infant born out of consanguineous marriage presented with respiratory distress for 2 weeks which worsened over last 3 days. He had significant history of diaphoresis, feeding difficulties and poor weight gain since birth. On examination, patient had tachypnoea, tachycardia, chest retractions, massive hepatomegaly, generalised hypotonia. Cardiovascular examination revealed loud S1 due to tachycardia, normal S2, no murmur. As the baby was extremely sick, emergency echocardiography was done. 2D echocardiography in parasternal long axis and short axis view revealed massive biventricular hypertrophy with slit like left ventricular cavity as if it is squeezed by thick ventricular wall (FIGURE 1A & 1B).



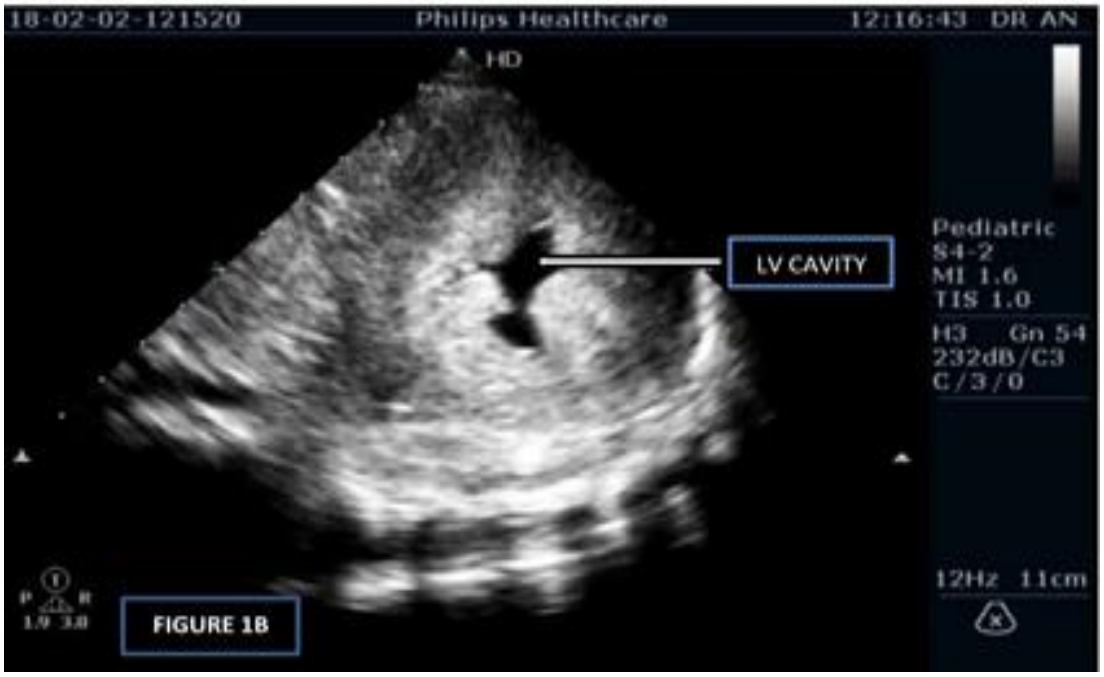
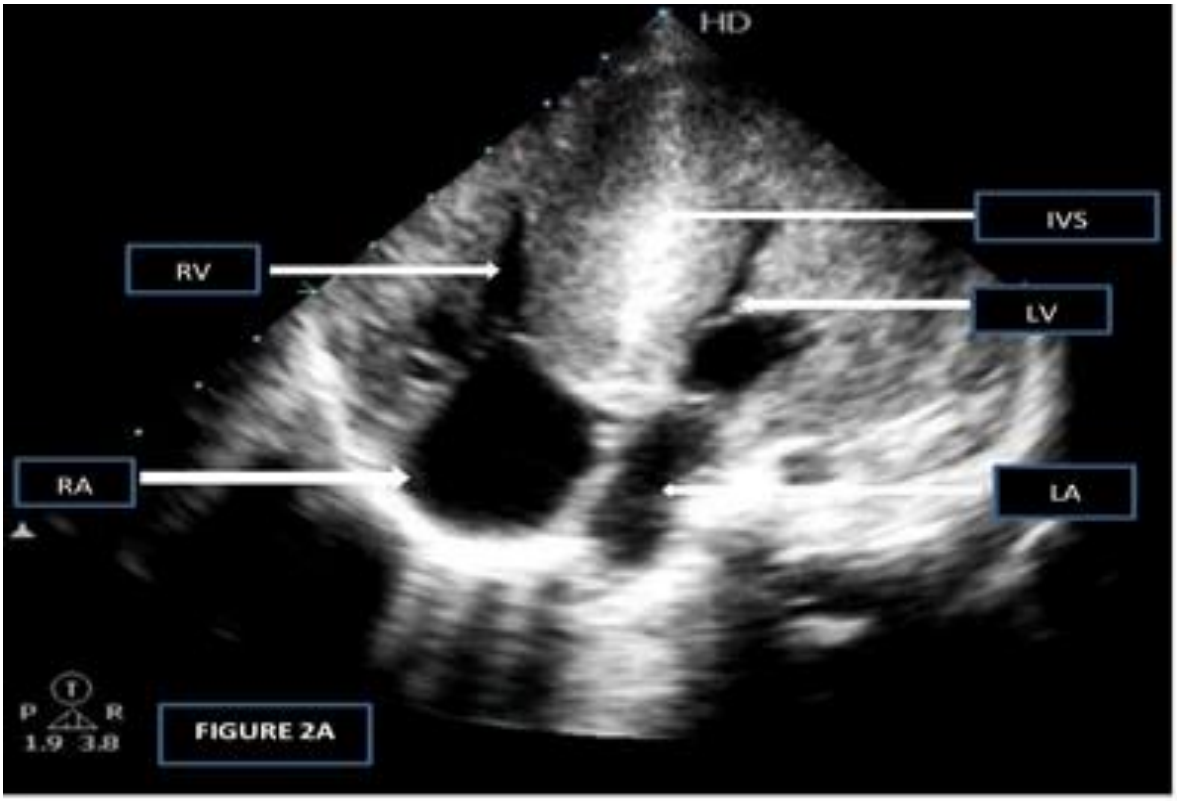


Figure 1A: *Parasternal long axis view showing severe concentric hypertrophy of left ventricle without left ventricular outflow tract obstruction. LV: Left ventricle, LVPW: Left ventricular posterior wall, IVS: Interventricular septum, LA: Left atrium.*

Figure 1B: *Parasternal short axis view showing marked biventricular hypertrophy, slit like left ventricular cavity with symmetric hypertrophy of LV. LV: Left ventricle.*

Apical four chamber view revealed biventricular hypertrophy with normal sized atria (FIGURE 2A). Apical five chamber view showed normal left ventricular outflow tract with normal aortic valve (FIGURE2B).



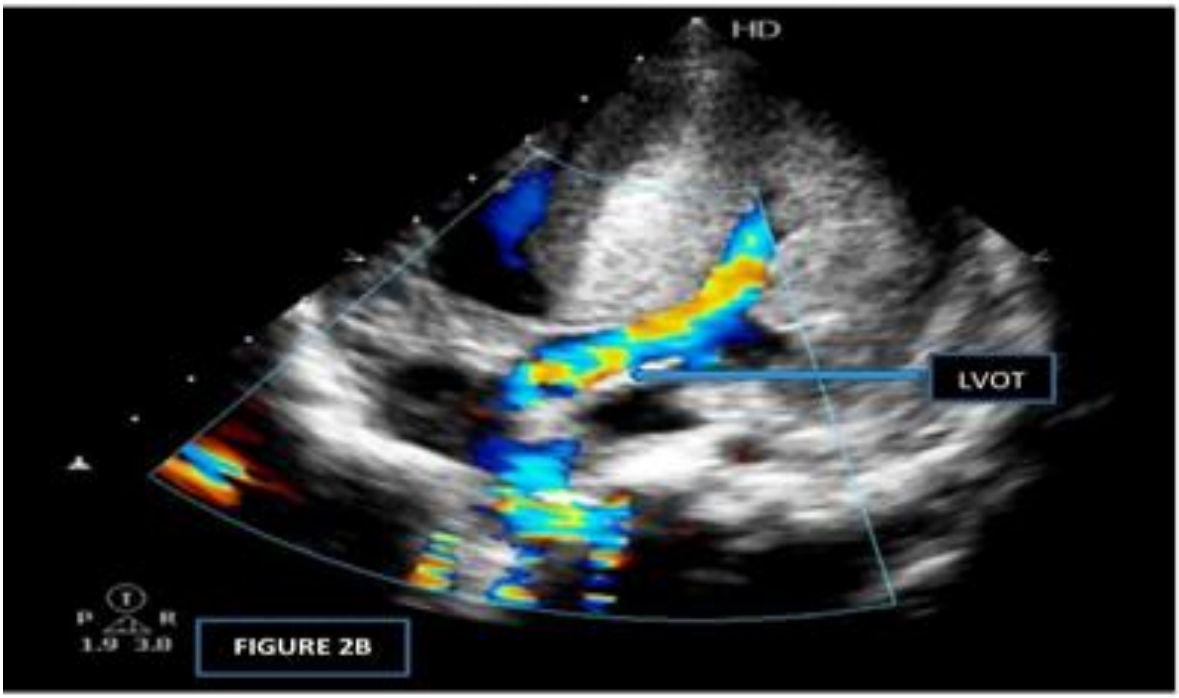


Figure 2A: *Apical 4-chamber view showing marked biventricular hypertrophy with thick interventricular septum. LA: Left atrium, LV: Left ventricle, RA: Right atrium, RV: Right ventricle, IVS: Interventricular septum.*

Figure 2B: *Apical 5-chamber view showing no flow turbulence across left ventricular outflow tract and no significant gradient. LVOT: Left ventricular outflow tract*

Subcostal view revealed normal sized atria with normal inter atrial septum (FIGURE 3A). Suprasternal view ruled out any evidence of coarctation of aorta (FIGURE 3B).

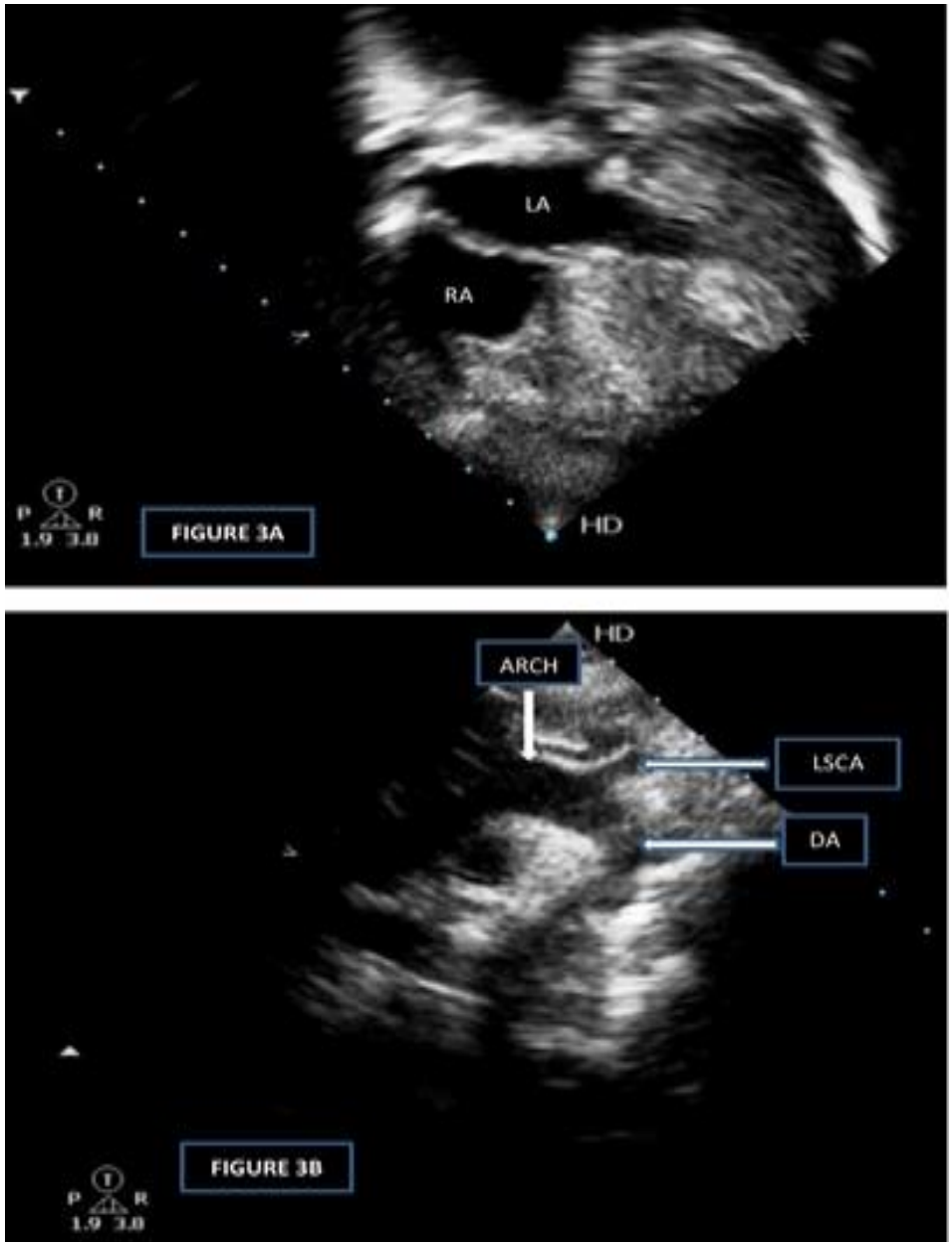


Figure 3A: *Subcostal view showing two atria with normal interatrial septum without any thickening. RA: right atrium, LA: Left atrium.*

Figure 3B: *Supratsernal view showing normal aorta and is branches. No evidence of coarctation was seen. ARCH: Arch of aorta, LSCA: Left subclavian artery, DA: Descending aorta*

In view of suspected diaphragmatic palsy patient was shifted to paediatric neurology ICU. Biventricular hypertrophy without left ventricular outflow tract (LVOT) obstruction was the provisional morphological diagnosis. Differential diagnosis of storage disorders (Type II, III or IV glycogen storage disease), HCM variants, PRKAG2 mutation, Danon disease, infant of diabetic mother was thought of. In view of high clinical suspicion of Pompe's disease, α-1, 4-glucosidase leucocyte enzyme assay was sent. The enzyme was significantly low: 1.76nmol/h/mg protein (normal range, 3.3–14.5 nmol/h/mg protein) confirming the diagnosis of Pompe's disease (Type II glycogen storage disease). Final diagnosis was infantile onset Pompe's disease presented with generalised muscle weakness, hepatomegaly, and cardiomyopathy with biventricular hypertrophy. Unfortunately, the infant died of respiratory failure before enzyme replacement therapy could be started.

Discussion:

Pompe's disease or type II glycogen storage disease occurs due to deficiency of lysosomal acid α-1, 4-glucosidase enzyme [3]. It results in lysosomal glycogen accumulation principally in cardiac, skeletal, and smooth muscle cells. Pompe's disease has autosomal recessive inheritance, so history of consanguineous marriage is very frequent as in our case and affected babies are usually males. Pompe's disease can be of two types: Infantile onset or juvenile adult onset. Infantile onset Pompe's disease presents with generalised hypotonia, hepatomegaly, hypertrophic cardiomyopathy and cardio-respiratory failure before first birthday [4]. Juvenile and adult-onset variants have less clinical severity, usual presentation is progressive proximal muscle weakness without cardiomyopathy. Differential diagnosis includes Danon disease (LAMP2

mutation), PRKAG2 mutation, Fabry's disease. Danon disease (LAMP2 mutation) is a lysosomal glycogen storage disease with normal acid maltose whereas acid maltose is essentially low in Pompe's disease [5]. Danon disease has X-linked dominant inheritance, childhood onset, presents with cardiomyopathy, skeletal myopathy, mental retardation [5]. Death occurs usually in 2nd to 3rd decade. PRKAG2 mutation has autosomal recessive inheritance, usual presentation is hypoglycaemia, cardiomyopathy and early death in infancy. Fabry's disease occurs due to alpha-galactosidase A deficiency [6]. It is a x-linked-recessive disease usually presents with renal, cardiac, nervous system and gastrointestinal involvement [6]. But usual clinical manifestation of Fabry's disease occurs at 30-45 years of age, it is not clinically apparent in infancy. In our case mother was nondiabetic. So maternal diabetes related ventricular hypertrophy of baby was ruled out. Infantile onset hypertrophic cardiomyopathy was also thought of which is exceedingly rare.

Successful treatment is available for Pompe's disease in the form of enzyme replacement therapy [Recombinant human GAA: alglucosidase alfa, (Myozyme)]. Timely administration of enzyme replacement can prevent fatality and regress organomegalies to a great extent. To conclude, in every case of floppy infant with hepatomegaly and biventricular hypertrophy without LVOT obstruction, Pompe's disease should be suspected, α-1, 4-glucosidase in leucocytes should be assessed as confirmatory test, prompt enzyme replacement therapy must be started if Pompe's disease is diagnosed.

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