Case Report

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Supraventricular Tachycardia as an Unusual Presentation of Infantile Onset Pompe Disease: A Case Report

Paediatrics Section

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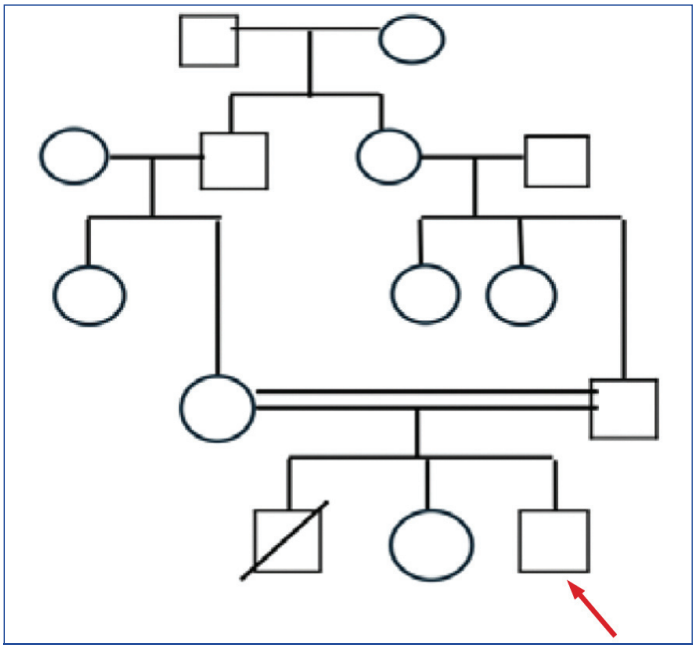
ABSTRACT

Pompe disease is a rare autosomal recessive lysosomal storage disorder with an incidence of approximately one in 40,000 live births is caused by deficiency of acid α-glucosidase, leading to progressive glycogen accumulation in cardiac and skeletal muscle. Infantile Onset Pompe Disease (IOPD) represents the most severe phenotype and usually presents within the first year of life with hypotonia and cardiomyopathy. An 11-month-old male infant third born of third-degree consanguinity, presented with irritability, lethargy, fast breathing and refusal of feeds. Clinical examination revealed a floppy infant with generalised hypotonia, proximal muscle weakness, absent deep tendon reflexes, with a peak heart rate of 280/min. Electrocardiography (ECG) revealed Supraventricular Tachycardia (SVT). SVT reverted with two doses of adenosine. Laboratory work-up of metabolic myopathies were within normal limits except for the Creatine Phosphokinase (CPK) which was elevated. Echocardiography revealed concentric left ventricular hypertrophy with severe systolic dysfunction. Genetic study identified a homozygous pathogenic variant in the GAA gene, confirming IOPD. The child was stabilised with anti-failure therapy and referred for enzyme replacement therapy. Unfortunately, infant succumbed in two weeks. Arrhythmias, including SVT, may be an early and intrinsic manifestation of the disease. Early recognition, genetic confirmation, vigilant cardiac monitoring, and timely initiation of enzyme replacement therapy are essential to improve survival and long-term outcomes in IOPD.

Keywords: Cardiomyopathy, Enzyme replacement therapy, Floppy infant, Glycogen storage disorder, Metabolic disorder

CASE REPORT

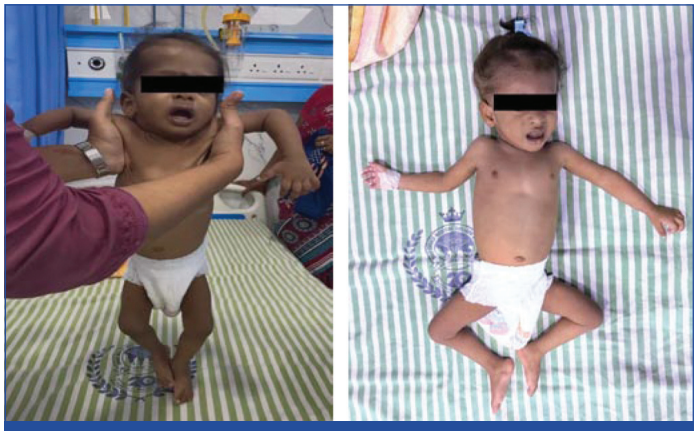
An 11-month-old male infant, third born to parents in a third-degree consanguineous [Table/Fig-1] marriage, presented to department of paediatrics with a three-day history of cough, cold, irritability, lethargy, fast breathing and refusal of feeds. There was no history of fever, seizures, vomiting, diarrhoea. Parents also reported poor weight gain and delayed motor milestones since early infancy. A family history revealed sudden unexplained death of an elder male sibling at 10 months of age. Child was fully immunised till nine months according to National Immunisation schedule. Child was on complementary feeds along with breastmilk.



[Table/Fig-1]: Pedigree chart, squares represent the males and circles represent the females showing a male child death (square with diagonal slash), normal female child (circle) and the index child (square indicated with arrow).

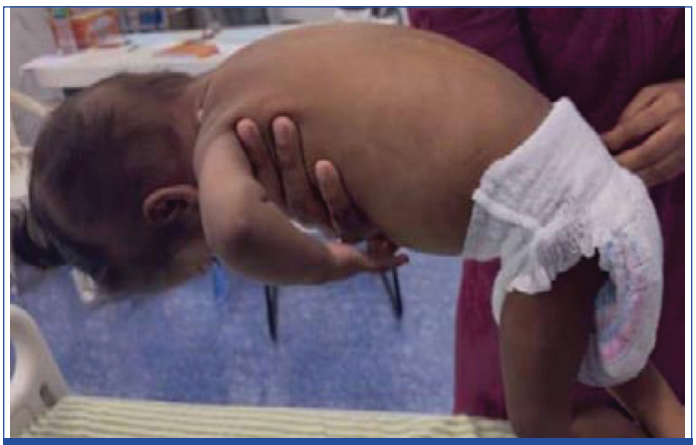
On examination, the infant was lethargic, tachypnoeic with respiratory rate of 62 breaths/min. Initial heart rate of 190/min, which subsequently increased to a peak rate of 280/min during the tachyarrhythmia episode. Capillary Refill Time (CRT) <2 sec, with saturation 96% under room air, normal blood pressure (72/58 mmHg), neurological examination revealed generalised hypotonia with a frog-leg posture. Muscle bulk was symmetrically reduced in all four limbs, with proximal weakness greater than distal weakness. Deep tendon reflexes were absent, plantar reflex were withdrawal in both legs. There was hepatomegaly with liver span of 9 cm [Table/Fig-2-4]. Anthropometry was suggestive of severe acute malnutrition (weight: 4.5 kg <3rd centile; length: 65 cm <3rd centile; mid upper arm circumference: 9 cm) with microcephaly (HC: 40.5 cm <-3 SD).

Baseline investigations showed haemoglobin 11.1 g/dL, which was lower limit of the normal for age, haematocrit 34%, RBC count 4.62 mill/cu mm, MCH 71.6 pg, MCV 24.1 fL, MCHC 32.6%, low T3, T4 and a low normal TSH, free T3 2.59 pg/mL, free T4 0.67 ng/dL, TSH 1.48 µIU/mL, possibly sick euthyroid state, mild elevation of potassium levels (pseudohyperkalaemia 5.3 mEq/L), due to



[Table/Fig-2]: Clinical image demonstrating axial hypotonia. (Images from left to right)

[Table/Fig-3]: Clinical photograph showing marked head lag on pull to sit examination.

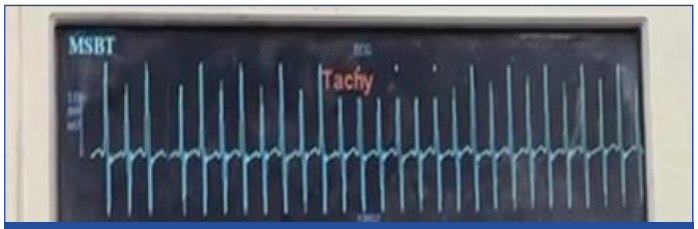


[Table/Fig-4]: Postural appearance suggestive of truncal and axial weakness.

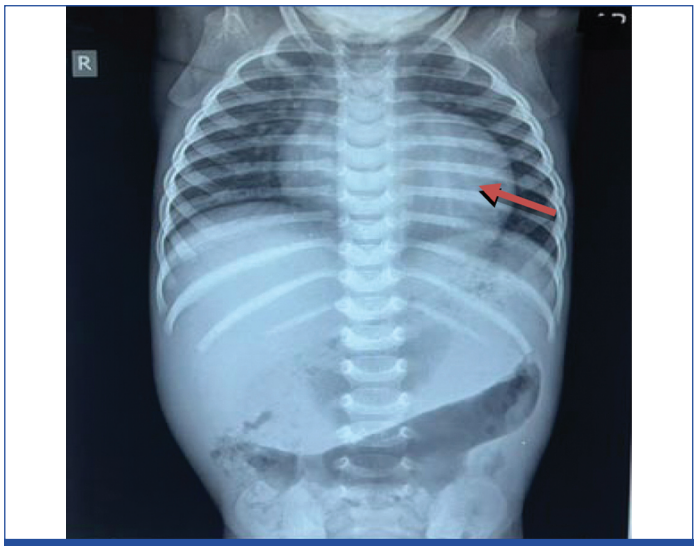
haemolysis as the supernatant of the centrifuged sample was pink, with the repeat value of 3.2 mEq/L, elevated SGOT and CPK levels [Table/Fig-5]. [Table/Fig-6] revealed narrow complex tachycardia with absent 'P' waves suggestive of SVT. Chest X-ray showed cardiomegaly [Table/Fig-7].

Tests	Observed value	Reference value
Complete blood count		
Haemoglobin (g/dL)	11.1	11.4-14.1
Haematocrit (%)	34	30-38
RBC count (mill/cumm)	4.62	3.9-5.1
MCH (pg)	71.6	72-84
MCV (fL)	24.1	25-29
MCHC (%)	32.6	32-36
TC ($\times10^9$/L)	7.23$\times10^9$	6-16$\times10^9$
Neutrophil (%)	39.3	15-45
Lymphocytes (%)	51.9	47-77
Monocytes (%)	6.2	2-10
Eosinophils (%)	2.1	1-6
Basophils (%)	0.5	0-0.6
Platelet ($\times10^3$/cumm)	358000	200-550$\times10^3$
CRP (mg/dL)	0.2	<0.6
Thyroid function test		
Free T3 (pg/mL)	2.59	2.9-6.8
Free T4 (ng/dL)	0.67	0.8-2.0
TSH (µIU/mL)	1.48	0.35-5.2
CPK (U/L)	**913**	**<200**
Renal function test		
Urea (mg/dL)	24	17.12-49.2
Creatinine (mg/dL)	0.56	0.04-0.59
Uric acid (mg/dL)	3.96	1.6-6.3
Sr. Sodium (mEq/L)	137	136-145
Sr. Potassium (mEq/L)	5.3 (Rpt: 3.2)	3.5-5.1
Sr. Chloride (mEq/L)	107	96-106
Sr. Ionised calcium (mEq/L)	1.19	1.15-1.29
Liver function test		
Total bilirubin (mg/dL)	0.46	0.2-1.2
Direct bilirubin (mg/dL)	0.16	0.05-0.30
Indirect bilirubin (mg/dL)	0.30	0.2-0.9
SGOT (Aspartate aminotransferase) (IU/L)	**274.3**	15-40
SGPT (Alanine aminotransferase) (IU/L)	**59.9**	15-40
Alkaline phosphatase (IU/L)	228.9	30-120
GGT (IU/L)	6.9	6-16
Total protein (g/dL)	6.35	6.0-7.8
Albumin (g/dL)	4.1	3.35-5.2
Globulin (g/dL)	2.25	2.5-3.5
A/G ratio	1.82	1.2:1-2:1

[Table/Fig-5]: Laboratory investigations show microcytic hypochromic anaemia, elevated Creatinine Phosphokinase and AST, TSH is low-normal with low Free T3 and Free T4 - suggests possible sick euthyroid syndrome, Potassium mild elevation noted with other parameters within normal range.



[Table/Fig-6]: Electrocardiogram showing narrow-complex tachycardia without visible P waves, suggestive of Supraventricular Tachycardia (SVT).

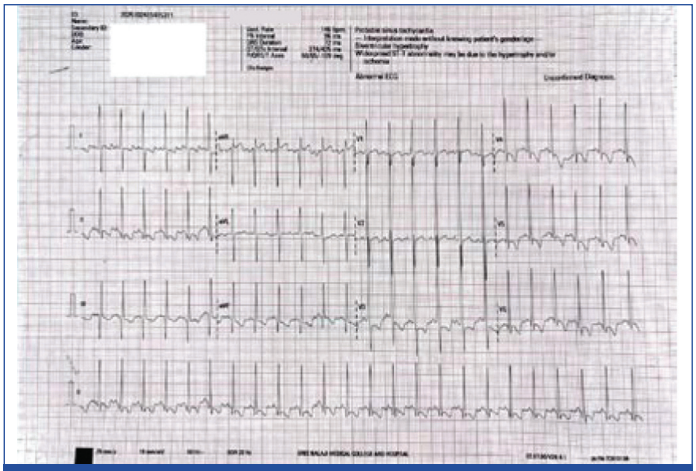


[Table/Fig-7]: Chest radiograph showed cardiomegaly with clear lung fields.

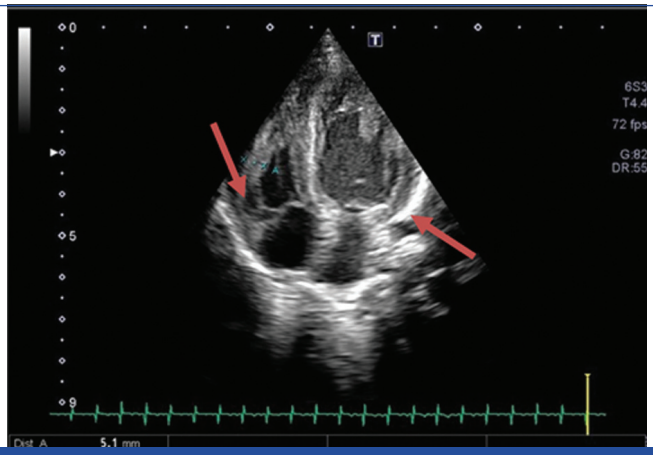
Electrocardiogram [Table/Fig-8] revealed sinus tachycardia with short PR interval and high-voltage QRS complexes (after reversion with adenosine). Echocardiography [Table/Fig-9,10] demonstrated concentric left ventricular hypertrophy with global hypokinesia and severe left ventricular systolic dysfunction (ejection fraction ~30%). Given the clinical suspicion of an inborn error of metabolism, genetic testing was performed.

Genetic Analysis [Table/Fig-11] showed whole-genome sequencing identified a homozygous pathogenic variant in the GAA gene, confirming IOPD, inherited in an autosomal recessive pattern.

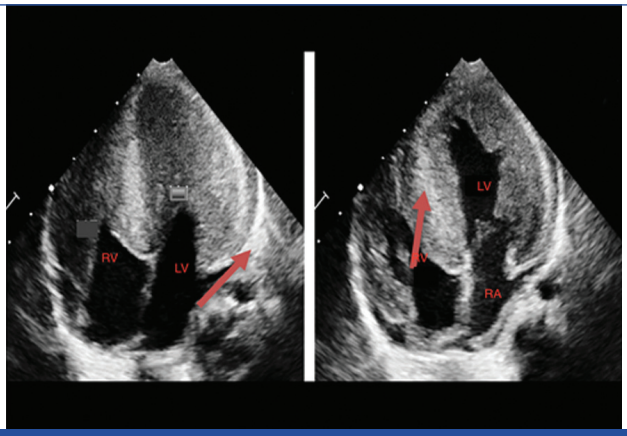
Differential diagnoses such as myocarditis, mitochondrial disorders, and other metabolic myopathies were considered. However, the lack of clinical features suggestive of an acute infectious process, along with



[Table/Fig-8]: Sinus tachycardia with short PR interval and high-voltage QRS complexes (after reversion with adenosine.



[Table/Fig-9]: Apical four-chamber view showing left ventricular hypertrophy (arrows); LVIDd 3.4 cm, LVIDs 3.0 cm.



[Table/Fig-10]: Echocardiographic images showing global LV hypokinesia with reduced systolic function; ejection fraction ~30% (arrows).

Test Results and Interpretation

HOMOZYGOUS PATHOGENIC VARIANT IN GAA GENE CONSISTENT WITH PHENOTYPE DETECTED : MOLECULAR DIAGNOSIS CONFIRMED.

Summary Of Variants

Gene and Transcript	Involved Exon-Intron/ Total Exon-Intron	Variant Nomenclature	Zygosity	Classification	OMIM Phenotype	Inheritance
GAA (NM_000152.5)	Exon 13/Exon 20	c.1798C>T p.Arg600Cys [244x /244x]	Homozygous	Pathogenic	Pompe disease, infantile-onset	Autosomal recessive

Variant Details

GAA

Variant Nomenclature	c.1798C>T (p.Arg600Cys)
Genomic Nomenclature	chr17:g.78086420C>T
Zygosity	Homozygous

Case Id: 51029614441 Patient Name: KAVIN

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[Table/Fig-11]: Genetic analysis showing a homozygous pathogenic GAA variant confirming Pompe disease.

the presence of generalised hypotonia, hypertrophic cardiomyopathy, and elevated creatine phosphokinase levels, made these less likely. The diagnosis was further supported by the identification of a homozygous pathogenic variant in the GAA gene, confirming IOPD.

SVT was reverted with two doses of adenosine (0.1 mg/kg followed by 0.2 mg/kg), resulting in reversion into sinus rhythm. Child was monitored in paediatric intensive care unit for three days. Following the stabilisation, child was initiated on anti-failure therapy with furosemide 1 mg/kg/day along with enalapril 0.1 mg/kg bd were initiated along with nutritional counselling. No recurrence of arrhythmias was noted during hospitalisation. Parents were counseled regarding the diagnosis, prognosis, recurrence risk and were also counseled regarding the need for family screening. The child was also started on propranolol 0.6 mg/kg/day and was referred to higher Centre for electrophysiological study and for initiation of enzyme replacement therapy with alglucosidase alfa.

Unfortunately, infant succumbed to illness within two weeks even before initiation of enzyme replacement therapy.

DISCUSSION

The IOPD is characterised by a consistent core phenotype of hypotonia and hypertrophic cardiomyopathy; Glycogen accumulation within cardiac myocytes and the specialised conduction system leads to myocardial hypertrophy and disruption of normal electrical pathways. This results in altered impulse generation and propagation, predisposing affected infants to a spectrum of arrhythmias, including supraventricular and ventricular tachyarrhythmias however, the clinical expression, severity, and cardiac rhythm manifestations show considerable heterogeneity [1], as evident when comparing previously published cases with the present institutional case report.

Martínez M et al., described a neonate with classic IOPD who manifested severe hypertrophic cardiomyopathy and developed recurrent episodes of ventricular fibrillation during central venous catheter placement, culminating in sudden cardiac death, thereby illustrating the extreme end of electrical instability associated with extensive myocardial glycogen infiltration [1]. In contrast, the present case presented at 11 months of age with established cardiomyopathy but survived the initial arrhythmic event, manifesting narrow-complex SVT rather than malignant ventricular arrhythmia. In the present case, SVT occurred in the absence of procedural or anaesthetic triggers, suggesting intrinsic electrical instability due to myocardial glycogen deposition. This expands the spectrum of arrhythmic manifestations in IOPD and highlights variability in clinical presentation. This difference suggests that while arrhythmogenesis is a shared feature, the type and lethality of arrhythmia may correlate with age at presentation, myocardial disease burden, and triggering factors, such as procedural stress in neonatal cases versus metabolic or infectious stress in older infants.

Starosta RT et al., in a case series, highlighted the occurrence of life-threatening arrhythmias during anaesthesia induction in infants with IOPD, emphasising procedural vulnerability in the setting of hypertrophic cardiomyopathy [2]. Unlike these cases, the arrhythmia in the present report occurred spontaneously during an acute illness, without exposure to anaesthesia or invasive procedures, underscoring that arrhythmias in IOPD are not limited to perianaesthetic settings. This finding is clinically significant, as it broadens the risk profile to include routine medical illnesses and reinforces the need for continuous cardiac surveillance even outside operative contexts. Furthermore, while Starosta RT et al., predominantly reported ventricular and bradyarrhythmia, the presence of SVT in the current case expands the arrhythmic spectrum associated with Pompe cardiomyopathy [2]. The pathophysiological basis of arrhythmogenesis in Pompe disease involves glycogen deposition in the conduction system, including the atrioventricular node and Purkinje fibers, leading to conduction abnormalities and susceptibility to tachyarrhythmias.

McDowell et al., evaluated arrhythmias in infants receiving Enzyme Replacement Therapy (ERT) and reported rhythm disturbances in approximately 18% of patients, raising concerns regarding both disease-related and treatment-associated electrophysiological instability [3]. In contrast, the arrhythmia in the present case preceded initiation of ERT, thereby supporting the concept that arrhythmias are an intrinsic manifestation of Pompe cardiomyopathy rather than solely a consequence of therapy. This distinction is crucial, as it emphasises the importance of early diagnosis and cardiac monitoring even before treatment initiation, particularly in resource-limited settings where delays in ERT initiation are common.

When compared with the present institutional case report, several classical features of IOPD were consistently observed across all reports, including hypotonia with lower motor neuron pattern weakness, elevated CPK levels, cardiomegaly, short PR interval, and high-voltage QRS complexes [4-7]. However, the present case is distinctive due to its combined presentation of SVT, severe left ventricular systolic dysfunction (ejection fraction ~30%), and survival beyond infancy, allowing for definitive genetic confirmation and referral for ERT. Additionally, the presence of third-degree consanguinity and a history of unexplained sibling death in the present case strongly support an inherited metabolic aetiology, aligning with established diagnostic guidelines and reinforcing the importance of family history in early recognition [8,9]. Differential diagnoses such as myocarditis, mitochondrial disorders, and other metabolic myopathies were considered; however, the presence of hypotonia, hypertrophic cardiomyopathy, elevated CPK levels, and confirmatory genetic findings supported the diagnosis of IOPD.

Overall, comparison with previously reported cases demonstrates that while cardiomyopathy is a universal finding in classic IOPD, arrhythmic manifestations vary widely in type, severity, and clinical context [3-6,10]. This case underscores the importance of considering metabolic aetiologies in infants presenting with unexplained cardiomyopathy and arrhythmias. Early recognition, genetic confirmation, and timely initiation of enzyme replacement therapy are critical in improving clinical outcomes.

The present case documented SVT as a presenting manifestation of IOPD, thereby expanding the recognised cardiac phenotype and highlighting the need to consider Pompe disease in infants presenting with unexplained tachyarrhythmia and cardiomyopathy. This comparative analysis reinforces in literature, the critical importance of early suspicion, genetic confirmation, vigilant cardiac monitoring, and timely initiation of enzyme replacement therapy to improve outcomes in affected infants [5,6,11]. Long-term outcome studies also demonstrate improved motor and cardiac function when therapy is initiated early in the disease course [12-14].

CONCLUSION(S)

Pompe disease should be strongly suspected in infants presenting with hypotonia and cardiomyopathy, particularly in the setting of consanguinity or a family history of unexplained infant deaths. Arrhythmias, including SVT, may be an early and intrinsic manifestation of the disease. Early recognition, genetic confirmation, vigilant cardiac monitoring, and timely initiation of enzyme replacement therapy are essential to improve survival and long-term outcomes in IOPD.

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