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Neonatal-Onset Primary Coenzyme Q10 Deficiency Type 5 with Multisystem Involvement: A Case Report

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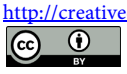

Abstract

Primary Coenzyme Q10 (CoQ10) Deficiency Type 5 is a rare, autosomal recessive mitochondrial disorder that impairs cellular energy production. Neonatal-onset forms often present with extensive multisystem involvement and carry a poor prognosis. CoQ10, a lipid-soluble component of the mitochondrial respiratory chain, plays a crucial role in oxidative phosphorylation. Mutations affecting genes such as *COQ*2, *COQ*4, or *PDSS*2 disrupt CoQ10 biosynthesis, leading to various mitochondrial pathologies. We report a case of a 3-month and 3-week-old Omani male diagnosed with genetically confirmed Primary CoQ10 Deficiency Type 5. The infant presented with intractable seizures from birth, lactic acidosis, global developmental delay, hypotonia, dilated cardiomyopathy, recurrent aspiration pneumonia, and failure to thrive. Despite early initiation of high-dose CoQ10 and comprehensive multidisciplinary care, the patient continued progressive neurological and metabolic decline. This case underscores the critical importance of early genetic diagnosis and highlights the limited therapeutic efficacy of CoQ10 supplementation in severe neonatal presentations. Coordinated, multidisciplinary management and family counseling remain essential to optimize care and guide expectations.

Subject Areas

Pediatrics

Keywords

Coenzyme Q10 Deficiency, Mitochondrial Disorder, Neonate, Seizures, Cardiomyopathy, Lactic Acidosis, Genetic Diagnosis, Multisystem Involvement

1. Case Report

A 3-month and 3-week-old Omani male infant was diagnosed with Primary Coenzyme Q10 (CoQ10) Deficiency Type 5, confirmed by whole exome sequencing. The patient was delivered at term after a pregnancy complicated by intrauterine growth restriction (IUGR). The mother, aged 41 years and gravida 7, para 4, living 2, abortion 2 (G7P4L2A2), had a history of two intrauterine fetal demises (IUFD) at 32 - 34 weeks and two spontaneous abortions at 8 weeks. Antenatal anomaly scans were normal, and no hypertension was reported during the pregnancy. Group B Streptococcus (GBS) and virology screenings were negative. At birth, the infant exhibited dysmorphic features and respiratory distress, requiring immediate admission to the neonatal intensive care unit (NICU). Seizure activity was noted from the first day of life. Electroencephalography (EEG) demonstrated severe multifocal epileptiform discharges with interictal suppression. Magnetic resonance imaging (MRI) revealed mega cisterna magna and subependymal cysts.

The infant exhibited persistent myoclonic and focal seizures refractory to treatment with levetiracetam, clonazepam, phenobarbital, and carbamazepine. Clinical examination revealed global developmental delay and hypotonia. Following genetic diagnosis, supplementation with CoQ10 (71 mg three times daily) and pyridoxal-5-phosphate was initiated. Brain MRI showed a large retrocerebellar space with a cerebrospinal fluid signal and a normal cerebellar vermis (**Figure 1**).

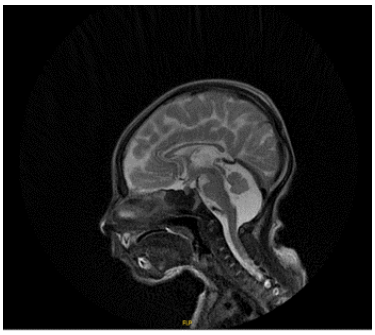

Figure 1. Brain MRI shows large retro cerebellar space that follows CSF signal on all sequences. Normal cerebellar vermis.

The patient experienced multiple respiratory infections, including influenza B and respiratory syncytial virus (RSV), requiring continuous positive airway pressure (CPAP), high-flow nasal cannula (HFNC), and home oxygen therapy. Recurrent aspiration pneumonias and chronic carbon dioxide retention led to multiple pediatric intensive care unit (PICU) admissions.

Echocardiography demonstrated progressive dilation of the left atrium and left ventricle with preserved systolic function, suggestive of early dilated cardiomyopathy (**Figure 2**). The aortic valve was functionally bicuspid (**Figure 3**). Frequent premature atrial and ventricular contractions were observed, likely secondary to metabolic instability. Medical management included captopril, spironolactone,

and hydrochlorothiazide.

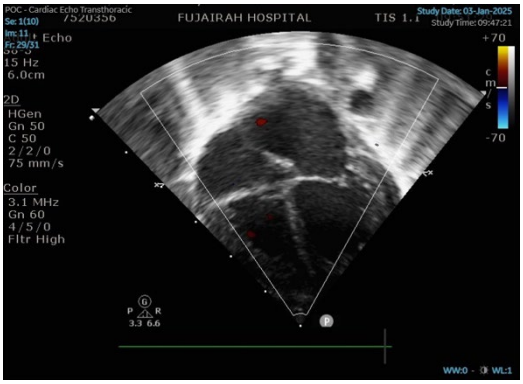

Figure 2. Apical 4 chambers view reveals LA and LV dilation.

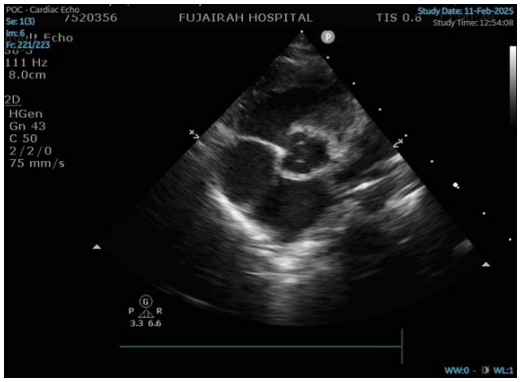

Figure 3. Para sternal short axis (PLAX) view reveals functionally bicuspid aortic valve.

Due to aspiration risk, oral feeding was discontinued. Gravity feeding of 90 mL every 4 hours was implemented, with gastrostomy tube placement under consideration. The patient was severely underweight (4.3 kg) and received iron and vitamin D supplementation. Laboratory studies revealed anemia (hemoglobin 8.1 g/dL) and hypoalbuminemia, both of which were treated accordingly.

Persistent lactic acidosis (serum lactate 6 - 9.5 mmol/L), elevated plasma amino acids (proline, alanine, glycine), and urine lactic aciduria and ketonuria were documented. Carnitine profiling showed an increased total-to-free carnitine ratio. Hyponatremia was managed with sodium supplementation. Renal and hepatic functions remained within normal limits.

2. Management and Outcome

The patient's treatment regimen incorporated a multidisciplinary strategy, addressing multiple systems: Neurological interventions included antiepileptic drugs such as levetiracetam, clonazepam, phenobarbital, and carbamazepine, alongside pyridoxal-5-phosphate and CoQ10 supplementation. Cardiac management involved captopril, spironolactone, and hydrochlorothiazide. Respiratory support consisted

of home oxygen therapy at 3 L/min and CPAP as needed. Infections were treated with intravenous ceftriaxone for pneumonia and appropriate antiviral agents. Nutritional support comprised iron and vitamin D supplementation, administered via gravity feeding. Supportive care encompassed physiotherapy, speech therapy, and genetic counseling to optimize overall patient management.

Despite comprehensive care, neurological impairment and metabolic instability persisted. Seizure control remained suboptimal, and developmental milestones were not achieved. The prognosis was considered poor, and palliative care options were discussed with the family.

3. Discussion

Primary Coenzyme Q10 Deficiency Type 5 (COQ10D5) is a rare autosomal recessive mitochondrial disorder caused by pathogenic variants in the COQ9 gene, which encodes a protein essential for the biosynthesis of Coenzyme Q10 (CoQ10). CoQ10 plays a central role in the mitochondrial respiratory chain and antioxidative defense, and its deficiency leads to impaired oxidative phosphorylation and multisystemic energy failure [1] [2]. Neonatal presentations, as in this case, often involve severe neurologic, cardiac, and metabolic derangements, and are associated with poor prognostic outcomes.

The patient's clinical presentation—refractory seizures, lactic acidosis, hypotonia, failure to thrive, and early-onset cardiomyopathy—was consistent with previously reported neonatal-onset COQ10D5 cases [3]-[5]. The persistent metabolic acidosis and elevated lactate reflect mitochondrial dysfunction, while the neuroimaging findings of mega cisterna magna and subependymal cysts may represent structural vulnerabilities resulting from in utero mitochondrial stress [6]. Neurological manifestations are particularly severe in COQ10D5 due to the high energy dependence of the central nervous system.

Although CoQ10 supplementation is the mainstay of therapy, particularly in primary CoQ10 biosynthetic defects, its efficacy is highly variable. A 2024 multicenter review of mitochondrial cytopathies highlighted that neonatal-onset patients, especially those with COQ9 mutations, showed minimal neurological improvement despite early and high-dose CoQ10 therapy [7]. Our patient, despite early supplementation and adjunctive pyridoxal-5-phosphate, demonstrated persistent seizures and global developmental arrest, supporting this observation. Moreover, epilepsy in CoQ10 deficiency is typically refractory to standard antiepileptics, as seen in our case, underscoring the need for future trials that target mitochondrial pathways more directly. [8].

Cardiac involvement, including early dilated cardiomyopathy and arrhythmias, is well-documented in mitochondrial disorders and has been reported in over 50% of COQ10D5 cases [9]. Our patient developed progressive atrial and ventricular dilation with preserved systolic function, managed conservatively. Though cardiac manifestations may sometimes respond to CoQ10, the progressive nature in this case underscores the aggressive disease phenotype.

Infectious complications and respiratory compromise, as encountered with recurrent pneumonia and carbon dioxide retention, further complicated management. Aspiration risk due to hypotonia and bulbar dysfunction is common and may justify early gastrostomy in similar cases. Nutritional support, although implemented, was insufficient to reverse severe failure to thrive—another hallmark of mitochondrial energy deficiency [10].

This case underscores the importance of early genetic diagnosis, especially when metabolic, neurological, and cardiac signs converge in neonates. Whole exome sequencing remains the gold standard in undiagnosed multisystemic presentations and facilitates targeted therapy initiation [11]. However, despite such interventions, the prognosis remains guarded. Neonatal-onset forms carry a high risk of mortality and severe neurodevelopmental outcomes, often justifying early discussion of palliative care options with families [12].

Future directions should focus on novel therapies, including gene replacement strategies and synthetic CoQ10 analogues with improved bioavailability. Early-stage trials using liposomal and nanoparticle-encapsulated CoQ10 compounds are currently under investigation and may offer hope for improved CNS penetration and efficacy in early-onset cases [13].

4. Conclusion

This case highlights the devastating clinical course of neonatal-onset Primary Coenzyme Q10 Deficiency Type 5 (COQ10D5), characterized by profound neurological, cardiac, and metabolic dysfunction. Despite early diagnosis via whole exome sequencing and prompt initiation of CoQ10 supplementation and multidisciplinary care, the patient's clinical condition remained severely compromised. The poor response to therapy underscores the aggressive nature of COQ10D5 in neonates and the current limitations of available treatments. This case emphasizes the importance of early genetic testing in infants with multisystemic involvement, the need for proactive family counseling, and consideration of palliative care in severe cases. Continued research into more effective therapeutic strategies, including next-generation CoQ10 formulations and gene-based interventions, remains critical to improving outcomes in this rare but devastating disorder.

Acknowledgments

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Ethical Approval

Signed informed consent was obtained from the patient's parents in accordance with the Ministry of health and prevention Research Ethics Committee (REC). This study was exempted from Ethical approval.

Conflicts of Interest

The authors declare no conflicts of interest.

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Abbreviations

CoQ10	Coenzyme Q10
NICU	Neonatal Intensive Care Unit
EEG	Electroencephalogram
MRI	Magnetic Resonance Imaging
IUGR	Intrauterine Growth Restriction
CPAP	Continuous Positive Airway Pressure
HFNC	High-Flow Nasal Cannula
PICU	Pediatric Intensive Care Unit
Hb	Hemoglobin
TID	Three times a day
IV	Intravenous
G-tube	Gastrostomy Tube
CNS	Central Nervous System
LPM	Liters per minute
LV	Left Ventricle
LA	Left Atrium