BIMDG

British Inherited Metabolic Disease Group

Leonardo Royal Hotel, Oxford

Pre-meeting
Monday 29 June 2026

Annual Symposium
Tuesday 30 June - Wednesday 1 July 2026

BIMDG Clinical & Scientific Trainees Session
Agenda
1 July 2026
9.00-11.00am
Location: Leonard Royal Hotel, Oxford

Time	Talk
09:00	**Introduction** Jennie Freestone
09:05	**An acute decompensation of Short-chain enoyl-CoA hydratase (ECHS1) deficiency** Nadia Amankrah
09:25	**A case series of sudden death in children aged 12 months to 4 years from previously undiagnosed inborn errors of metabolism** Sophie Manoy
09:45	**A case with many missed clues** Corey Pritchard
10:05	**Metabolic quiz and cases** A fun and informative Inborn Errors of Metabolism quiz involving patient cases, and using the interactive software Mentimeter

Presentation Abstracts

An acute decompensation of Short-chain enoyl-CoA hydratase (ECHS1) deficiency

Dr Nadia Amankrah, Dr Emily Sivers,
University Hospitals of Leicester NHS Trust

A 20-month-old male presented to Paediatric Emergency Department (PED) with 1 day of cough. He had a background of mitochondrial Short-chain enoyl-CoA hydratase (ECHS1) deficiency, diagnosed at 14 months. Though he was yet to receive an Emergency Care Plan, parents were advised during clinic appointments with the Metabolic Team that he required lactate and ammonia review during acute presentations. Observations, venous blood gas (including lactate) and ammonia were unremarkable, and he was discharged.

He returned 24 hours later with increased work of breathing. On initial observations, he was tachycardic (156bpm) and tachypnoeic (155bpm). He was managed with inhaled bronchodilators, but within 2 hours, had become dystonic. Capillary blood gas (CBG) showed metabolic acidosis (pH 7.236, HCO_3^- 10.9mmol/L, PCO_2 1.89kPa, lactate 3.0 mmol/L, anion gap 22.1 mmol/L). Ammonia was unremarkable (15µmol/L). Several attempts at intravenous (IV) and intraosseous cannulation were made to administer fluids. Repeat CBGs showed severe metabolic acidosis (pH 6.937), and he ultimately required a central venous catheter, intubation and admission to Children's Intensive Care Unit.

His metabolic acidosis was managed with IV sodium bicarbonate, 2 days after which his blood gas ptimized. Magnetic Resonance Imaging (MRI) of the head showed progression of previously established neurodegeneration in basal ganglia and cerebellum. Electroencephalogram was normal. He was stepped down to a General Paediatric Ward 5 days later,

where he received input from the Neurology Team for ongoing dystonia, and Chest Physiotherapists for respiratory secretions.

He was discharged approximately 40 days after presentation on Co-enzyme Q10 and several anti-dystonic medications, with new requirements for nasogastric feeding and airway suctioning at home. This case of decompensation of a rare neurometabolic disorder highlights the potential for rapid deterioration and importance of early blood gas in acute presentations of patients with ECHS1 deficiency, particularly where initial lactate and ammonia levels are not markedly elevated.

A case series of sudden death in children aged 12 months to 4 years from previously undiagnosed inborn errors of metabolism

Dr Sophie Manoy[1, 3, 4], Sally Smith[1, 2], Dr Marita Smith[1], Catherine Atthow[1], Janette Spicer[1], Dr Matthew Lynch[1, 4], Dr Michelle Lipke[1], Dr Julie A McEniery[4,7,8], Prof David Coman[1, 4, 5], Anita Inwood[1, 2], Dr Jim McGill[6], Dr Carolyn Bursle[1]
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Background: Sudden death in children aged 12 months to 4 years is rare. Awareness of causes from inborn errors of metabolism (IEM) can aid in rapidly identifying life-threatening biochemical derangements for immediate treatment. LPIN1 deficiency and inorganic pyrophosphatase 2 (PPA2) deficiency are recently described IEM that can result in unexpected death from sudden cardiac arrest. Recognition of patterns of acute presentation can raise clinical suspicion of these disorders, helping to ensure that appropriate investigations and sub-specialist consultation occur to optimize intervention, provide diagnostic certainty and facilitate genetic counselling for families.

Method: We reviewed the records of the Queensland Lifespan Metabolic Medicine Service (QLMMS) to identify children who experienced sudden cardiac arrest with subsequent death without prior known diagnosis of IEM, aged between 12 months and 4 years of age during 2010 to 2025. We aimed to describe patterns of presentation including clinical signs and laboratory findings to improve diagnosis recognition.

Results: Five children aged between 12 months and 4 years were included. There were three cases with LPIN1 deficiency, average age of death being 3 years, and two cases with PPA2 deficiency, average age at death being 22 months. Most (80%) did not have medical co-morbidities and presented with a rapidly progressive deterioration in the setting of viral-like illness. Where medical history was present (40%), there were features suspicious of these conditions (myoglobinuria, cardiac arrhythmia).

Conclusion: LPIN1 deficiency and PPA2 deficiency are IEM that can cause sudden death in this age group. Presenting features include severe rhabdomyolysis, refractory hyperkalaemia or sudden cardiac arrhythmia/arrest. Recognition will allow appropriate biospecimen collection including potassium, creatinine kinase, urine myoglobin, and DNA for molecular testing. Diagnosis can lead to potentially life-saving intervention for children and siblings, facilitate diagnostic certainty and provide options for future family planning with appropriate genetic counselling in this unfortunate scenario.

A case with many missed clues

Corey Pritchard, Dr Vicki Warburton, Rebecca Hopkins
University Hospitals Bristol and Weston NHS Trust

This is a case of a 44-year-old male who initially presented in 2007 with pigmented sclera. A referral was made to Ophthalmology who diagnosed benign pigmentation of pingueculae. He was monitored for years with no change so was discharged. He wasn't seen again by Ophthalmology until December 2025. On this occasion the clinical opinion was this was not benign pigmentation of pingueculae and a referral was made to Liverpool Ocular Oncology Centre. On review the team suspected Ochronosis.

A urine sample for organic acid analysis showed gross excretion of homogentisic acid, consistent with a diagnosis of Alkaptonuria. The urine had a creatinine of 0.4 mmol/L by Beckmann Enzymatic creatinine assay, unusually dilute for an adult patient.

On review of the patient's clinical history, in 2007 he had presented with renal stones and osteoarthritis in his left shoulder. He had a shoulder replacement and recalled the orthopaedic surgeon commenting that "his bones were black". At this time, renal stone analysis had been performed and observations of black stones made. No common renal stone constituents were detected. Urine oxalate was also elevated on a sample with creatinine of 0.51 mmol/L.

The low urine creatinine results raised suspicion of reliability. Interference of homogentisic acid in routine biochemistry investigations, specifically Trinder reaction assays, was found to be reported in the literature. He had regular urea and electrolytes due to a diagnosis in 2022 of clear cell renal cell carcinoma and several episodes of acute illness described as "atypical AKI". Both urine and plasma samples were referred to UHW Cardiff for creatinine by the Jaffe method. Results were notably higher by the Jaffe method. An alert was added to the patient record to flag this interference issue for subsequent requests. The patient has since been referred to the Adult Metabolic Service at UHBW.