Abstracts

reduced alpha-glucosidase activity but had normal urine glucose tetrasaccharide excretion; one was compound heterozygote for variants of unknown significance (VUS) and the other had no clear pathogenic mutations. The remaining case had their blood spot alpha-glucosidase activity repeated which was towards the bottom of the reference interval, but urine tetrasaccharide excretion was normal. All 5 patients remain healthy on follow-up.

Case	Age at referral (months)	Bloodspot Enzyme Activity	Lymphocyte Enzyme Activity	Urine Glucose tetrasaccharide (HEX4)	GAA gene analysis
1	4.6	Normal	Marginally reduced	< 5 umol/L	2x VUS
2	3.5	Low end of normal range	Not done	< 5 umol/L	Not done
3	2.4	Not done	Marginally reduced	< 5 umol/L	No pathogenic mutations detected
4	2.7	Not done	Normal	Not done	Not done
5	8.0	Normal	Normal	Not done	Not done

VUS: variant of unknown significance

Conclusions Our experience of the follow-up of newborn screen-positive Pompe disease corroborates the high false positive reported by Pompe disease screening programs around the world. Since the UK does not screen for Pompe disease we developed a local confirmatory diagnostic pathway for the investigation of these asymptomatic infants. While we conclude that the results of NBS for Pompe disease should be interpreted cautiously, a clear algorithm is required for the rapid identification of true positives to expedite management decisions, particularly prompt treatment with ERT. A balanced approach is required when counselling families to reduce unnecessary parental stress caused by a false positive result.

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DOES THIS CHILD HAVE COVID-19 – A DESCRIPTIVE OBSERVATIONAL STUDY AND EDUCATIONAL STRATEGY FROM A PAEDIATRIC EMERGENCY DEPARTMENT

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Background When the WHO declared a global pandemic on 12 March 2020, as emergency paediatricians we prepared for an influx of cases similar to adult services. The picture that evolved was different with a dramatic reduction in paediatric emergency department (ED) attendances in the UK. While adults presented with severe respiratory illness children were mainly identified following universal screening of admitted patients and presented innocuously with a febrile illness, cough and tonsillitis or lethargy with poor feeding in

neonates. These children needed either symptomatic treatment or a short period of admission, usually less than 48 hours and had an uncomplicated clinical course. In April 2020 a new condition associated with COVID-19, PIMS-TS emerged with a clinical picture ranging from the benign to the life-threatening making identification challenging for Emergency and Paediatric staff.

Objectives The aim was to enable general and acute paediatricians in identification, initial investigation and management of children with potential SARS-CoV-2 infection and PIMS-TS.

Methods A descriptive analysis paper using local BHRUT data and incorporating national guidelines was produced for front-line ED clinicians and paediatricians for appropriate identification of children with PIMS-TS who presented to ED. Good clinical examination was emphasised, review of observations and treating unwell children according to National Institute for Health and Care Excellence and Advanced Paediatric Life Support guidelines with early escalation and discussion with tertiary centres.

Results On review of local data it emerged that the first four children with PIMS-TS had already presented to ED at BHRUT in April 2020 before the RCPCH defined the condition. Three children were admitted based on the clinical acumen of the receiving clinician. The children presented with fluid refractory shock or cardiac dysfunction or partial Kawasaki features of rash and conjunctivitis. A 15 year old appeared to be well whilst having abnormal observation and had a tonsillar focus. She was eating and drinking but hypotensive at presentation. She went on to develop fluid refractory shock within 6 hours and was transferred to PICU but ultimately had a good outcome. A fourth child was picked up on the third attendance to ED with fluid refractory shock. The timeline of presentation of both children was plotted on a template and used as an educational strategy to emphasise the varied and heterogeneous presentation of PIMS-TS.

Conclusions Subsequently 22 cases of PIMS-TS presented to BHRUT. Then intensive education and raised alerts resulted in appropriate identification of these children. For the emergency paediatrician, the mainstay of management remains considering the diagnosis and instigating supportive measures for children with PIMS-TS with early involvement of specialist teams. Observational studies using index cases ensures that there is a high index of suspicion to inform medical and nursing staff who will see these children in the early stage of disease when then can present non specifically mimicking an endemic viral illness. Emphasis was also laid on the fact that the proportion of febrile children without serious pathology presenting to ED would remain considerably higher than those with PIMS-TS to prevent potential over investigation.

Paediatricians with Expertise in Cardiology Special Interest Group

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A RARE CAUSE OF LONG QT- TIMOTHY SYNDROME

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Background TS is an extremely rare genetic disorder of the L-type cardiac channel Ca(V)1.2 encoded by *CACNA1C*. The syndrome is characterized by multi-system abnormalities

consisting of QT prolongation, congenital heart defects, syndactyly, facial dysmorphism, and developmental delay and autistic spectrum disorder.

Timothy syndrome (TS) is a rare genetic disorder characterized by an abnormally prolonged cardiac repolarization time (long QT interval). This predisposes individuals to arrhythmias, cardiac arrest and sudden death.

Objectives We want to report a case of Timothy syndrome, incidentally detected during induction for general anaesthesia.

Methods Electronic records were used to collect data

Results An 8 yr old boy was admitted to hospital for elective orchidopexy, during induction he was developed 2-degree AV block with T alternans, maintaining reasonable cardiac output throughout. Past medical history of one admission with possible seizure. Of note he is currently being evaluated for autism.

He was born at 38 weeks. labour was induced due to poor growth.

His physical exam was normal He subsequently had a 12-lead ECG which showed a prolonged QTc 0.504s. His genetic testing shows a pathogenic gene mutation in CACNA1C. Parents have been counselled for the need for implantable defibrillator. He has been given an external automated defibrillator in the meantime.

He is currently on nadolol 40 mg OD. His parents are waiting gene testing.

Discussion Classic Timothy syndrome (TS) is a rare genetic disorder with dysfunction in multiple organ systems, clinically characterized by long QT syndrome and syndactyly. Timothy syndrome was first described in 1992 as sporadic cases of long QT syndrome, congenital heart disease and syndactyly. Since then rare cases have been reported in the literature. Classic TS is caused by a single missense mutation G406R of exon 8A of the Cav1.2 L-type calcium channel gene (CACNA1C) and is inherited in an autosomal dominant fashion, although it usually is the result of a de novo mutation. Patients with TS are prone to life threatening ventricular arrhythmias as a consequence of a prolonged QT interval. Other cardiac manifestations include septal defects, patent ductus arteriosus, cardiomyopathy and Tetralogy of Fallot.

Since the affected gene is widely expressed in multiple adult and foetal tissues including gastrointestinal system, brain, lungs, immune system and testes, extracardiac manifestations are common in patients with TS. Many present with developmental delay, cognitive abnormalities and autism.

These patients are at high risk for sudden death due to life threatening ventricular tachyarrhythmia. Implantation of an ICD at a very young age may be the best means to prevent sudden death.

Conclusions Timothy syndrome is a rare congenital arrhythmia disorder with dysfunction in multiple organ systems

The risk for life-threatening ventricular tachyarrhythmia is the limiting factor of TS. Since ventricular tachyarrhythmia is the leading cause of death in patients with TS, effective anti-arrhythmic medication and an implantable cardioverter defibrillator are the mainstay of therapy

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EFFICACY AND ADVERSE EFFECTS OF IBUPROFEN IN THE TREATMENT OF HAEMODYNAMICALLY SIGNIFICANT PATENT DUCTUS ARTERIOSUS (PDA) IN PRETERM INFANTS

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Background Ibuprofen is used as first line therapy to close haemodynamically significant patent ductus arteriosus (PDA) but is associated with severe gastrointestinal and renal effects.

Objectives The aims of this study were to determine the efficacy and safety of ibuprofen for pharmacological closure of PDA in preterm infants ≤ 29 weeks gestation.

Methods Retrospective study between 2015–2019 at a tertiary centre. 82 infants ≤ 29 weeks gestation with a haemodynamically significant large PDA on echocardiogram were enrolled. Data was collected using an electronic patient record system.

Results As shown in table 1, cohort had a median gestational age of 25+3 (range: 23+2 to 29+1), a median birth weight of 0.79kg (range: 0.39–1.25kg) and a median PDA diameter of 2.7mm (range 1.6–4.0mm). 70/82 infants completed their first course of ibuprofen, achieving full PDA closure in 25 patients (30.5%), partial closure in 29 (35.4%), and no closure in 24 (29.3%). 13/82 patients (15.9%) required a second course of ibuprofen, achieving full PDA closure in 3 patients (23.1%), partial closure in 1 (7.7%), and no closure in 9 (69.2%).

Associated adverse effects were reported in 13/82 patients (15.9%). These were mainly gastrointestinal and included necrotising enterocolitis in 5/82, bowel perforation in 5/82, and a further 1/82 had an upper gastrointestinal bleed. 9 patients subsequently underwent surgical ligation of their PDA, and an additional 5 patients died prior to PDA ligation due to non-cardiac complications.

Conclusions In our cohort ibuprofen was moderately efficacious in closure of PDA. Ibuprofen was generally well tolerated, although 13/82 infants (15.9%) were noted to have adverse effects. Whether these gastrointestinal adverse effects

Abstract 597 Table 1 Baseline characteristics of cohort

Baseline characteristics	Cohort no. = 82
Gestational age (weeks+days)	25 + 3 (range: 23+2 - 29+1)
Birth weight (kg)	0.79 (range: 0.39 - 1.25)
PDA diameter (mm)	2.7 (range: 1.6 - 4)
LA:Ao ratio	1.8 (range: 1.2 - 2.7)
Left to right shunt (n)	79 (96.3%)
Pulsatile flow pattern (n)	53 (64.6%)
Diastolic steal documented (n)	34 (41.4%)