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

Caroline Ponmani, Caroline Ponmani, Yvette Redpath, Sherin Koshy, Lunik Sarder, Gaurav Baruah, Sai Win, Tom Owens. ¹BHRUT; ²Barking Havering and Redbridge NHS Trust

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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 heterogenous 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

Anupama Mallappa, Detlev Rogahan. Royal Aberdeen Children's hospital

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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