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	Clinic	Feature in LOINC	Diagnosis	Genes in LOINC
SDS	Delayed weight gain	LP36298-5 Failure to thrive	Pancreas insufficiency	41764-2SDS gene-targeted mutation analysis in Blood or
SDS	Fatty stool	16142-2 : Fat [Mass/time] in 24 hour Stool	Pancreas insufficiency	Tissue by Molecular genetics method
SDS	Recurrent infections	751-8 Neutrophils [#/volume] in Blood by Automated count	Neutropenia	
Thal	Pale	718-7 Haemoglobine in blood	Anaemia	55234-9Alpha thalassemia gene panel - Blood by Molecular genetics method
Thal	Screening Newborns	46740-7 Haemoglobine disorders newborn screen interpretation	Hemoglobinopathy	

pregnancy, birth, growth, development, physical examination and vaccination, to promote and maintain health. We identified, among other codes, the *Logical Observation Identifiers Names and Codes*, a universal standard and electronic database for clinical care and management, applicable to child health care (such as codes 8339-4Birth weight Measured and 8287-5Head Occipital-frontal circumference by Tape measure) for registering the first feature of a (rare) disabling condition. Medical guidelines on Shwachman Diamond Syndrome (SDS) and Thalassemia Major were reviewed on recommended measurements. LOINC codes specific for SDS and thalassemia were applied in text mining for processing PubMed document sets.

Results We identified a subset of international interoperable codes to help to identify (rare) disabling conditions presenting in the first years of life. Child health handbooks can be enriched with a simple laboratory test to help diagnose diseases as a possible part of a (rare) condition.

Text mining is a powerful tool for processing PubMed document sets to identify diagnostic test in literature. Using unsupervised techniques such as clustering and spatial placement, with one can quickly gain insight into the contents of the documents, discover hidden properties and determine how to further explore and label the data.² Terms that belong to LOINC codes in filtering the whole (ranked) document set one can identify also the important rare disease papers that most likely are relevant for a medical test.

Conclusions The application of specific code-sets ensures the harmonization of data and the possibility of data exchange. As demonstrated with the LOINC the establishment of an interoperable child health record, including children with chronic illness and disabilities, is feasible. Collaboration between paediatricians, families, health system managers and data services is necessary to provide digital solutions to support the SDG3.

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IMPROVING PAEDIATRIC EMERGENCY DEPARTMENT REFERRALS TO FRACTURE CLINIC WITH THE INTRODUCTION OF A MUSCULOSKELETAL CLINIC – A RE-AUDIT

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Background Fracture clinic (FC) and Paediatric Emergency Department (PED) have a crucial relationship in the management of musculoskeletal (MSK) injuries in children. A high volume of patients are assessed in FC and even a small number of inappropriate and non-specific referrals severely disrupts the flow of FCs.

Chelsea and Westminster Hospital, London, has seen a rapid increase in children attending PED and subsequent referrals to FC. In light of this, we set up a MSK clinic.

Objectives To reduce the number of inappropriate referrals made from the PED to FC.

Methods PED referrals to FC prior to the introduction of the MSK clinic were audited. Data was collected retrospectively from 22 consecutive FCs over a 10-week period from February 2019 to April 2019. Patient demographics, diagnosis and management were recorded for these patients. PED referrals to FC were re-audited after the MSK clinic was set up. 18 consecutive FCs were retrospectively analysed over a 9-week period from January 2020 to March 2020. The appropriateness of these referrals was compared before and after the establishment of the MSK clinic.

An appropriate referral was defined as one in which the resulting diagnosis was a fracture, and a referral was classed inappropriate if the diagnosis was not.

Results The initial audit in 2019 which included 203 children referred to FC by PED showed that 35.5% of referrals were inappropriate. After the introduction of the MSK clinic, the re-audit demonstrated an improvement, as of the 178 referrals, only 26% were non-fractures.

Most notably, the number of 'unspecified injury' referrals decreased in 2020 compared to 2019 (29% in 2019 to 9% in 2020).

Conclusions The introduction of the MSK clinic has proven an effective way of reducing the number of inappropriate and non-specific referrals made by PED to FC.

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PRESCRIPTION OF PARACETAMOL AS AN ANTIPYRETIC IN PAEDIATRICS: ANALYSIS OF PRACTICES IN A NATIONAL ACUTE AND TEACHING HOSPITAL

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Background Prescription errors represent a pervasive problem found across many hospitals and the ubiquity by which antipyretics are prescribed in paediatrics makes them a frequent source of error. Such avoidable errors not only lead to actual physical harm for the child, but also incur financial and legal costs on the service provider, dampen public confidence in the health care system and predispose to negative psychological effects on both the patient and prescriber.

Objectives Prescriptions of Paracetamol for paediatric inpatients at Mater Dei Hospital, a national acute and teaching hospital

in Malta, were analysed for sources of error. The British National Formulary for Children (BNFC) was used to establish the correct prescribing standard.

Methods Treatment charts of all admissions to medical paediatric inpatient wards were reviewed daily over a four-week period. Prescriptions for Paracetamol were assessed for legibility, inedibility, approved drug nomenclature, correct dose and dosing frequency, approved dosing interval abbreviations, writing of minimum dosing interval for pro re nata (PRN), appropriate dating, prescriber signature and prescriber designation. Treatment charts were also analysed to assess accurate writing of patient name, identification number, age, date of birth, height, weight, and allergies. Paracetamol prescriptions for indications other than fever were excluded.

Results A total of 72 treatment charts were analysed of which 44 contained Paracetamol prescriptions. Age ranged from 1 day to 13 years. 93.2% of all prescriptions were on a PRN basis. Legibility and inedibility met the BNFC standard in 100% of cases. Approved drug nomenclature was used in 97.7% of prescriptions.

With regards to dosing, 54.5% of prescriptions did not follow the standard leading to incorrect dosing. Of these cases, 50% were due to the same dose of Paracetamol being prescribed for the oral, intravenous and rectal routes used for the same child. In the other 50%, the oral dose was calculated by weight instead of using fixed dose ranges based on age. Where errors were made, patients were overdosed by an average of 20% more than the recommended maximum dose. Correct dosing frequency was present in 100% of cases. 97.7% of dosing interval abbreviations were not according to guidelines, mainly because English abbreviations were not written in full. PRN was not written in 51.2% of Paracetamol PRN prescriptions.

Dating was correct in 95.5% of cases. Prescriber signature present in 97.7% of cases and prescriber designation in 95.5% of prescriptions.

The following table illustrates data related to treatment charts:

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Patient Criteria	Percentage Correctly Completed
Name	94.4
Identification Number	94.4
Age	83.3
Birth date	22.2
Height	1.4
Weight	76.4
Allergies	54.2

Conclusions Adherence to proper prescription practices has been repeatedly emphasised by regulatory bodies and safe prescription is considered an integral part of sound medical practice. Greater attention needs to be paid to appropriate dosing according to indication and route to prevent overdosing. Correct writing of dosing interval abbreviations, as well as recording of weight and allergies in treatment charts is also essential.

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MIND THE GAP!! UNCOMMON CAUSE OF HIGH ANION GAP METABOLIC ACIDOSIS DUE TO PYROGLUTAMIC ACIDOSIS (PGA)

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Background Metabolic acidosis is common in sick children. Measurement of anion gap helps to narrow down the cause of high anion gap metabolic acidosis such as DKA/lactic acidosis/renal failure. A rare cause of high anion gap metabolic acidosis is due to accumulation of pyroglutamic acid. Acquired PGA occurs in association with glutathione and cysteine depletion or as a side effect of certain medications. We report a complex case of child with GLIS3 mutation who had high anion gap metabolic acidosis secondary to acquired pyroglutamic aciduria associated with common drugs such as paracetamol.

Objectives

Methods

Results 10 yrs old girl was born at 35 weeks gestation with severe growth retardation with birth weight of 1.17 kg. She developed neonatal diabetes on day 3 and congenital hypothyroidism on day 4. The combination of neonatal diabetes, congenital hypothyroidism and some dysmorphic facial features led to a clinical diagnosis of a GLIS 3 mutation, subsequently confirmed by genetic analysis. Her neonatal diabetes was managed with insulin pump therapy and high dose thyroxine in order to maintain her thyroid function. She was treated for exocrine pancreatic insufficiency, gastro oesophageal reflux with jejunostomy feeds. She also had liver transplantation at the age of 3 yrs for hepatic fibrosis and cirrhosis. She had polycystic kidney disease and deteriorating renal function, hypertension, chronic anaemia, recurrent chest infections and developmental delay. She was admitted with a viral illness and chest infection treated with cefuroxime, paracetamol and salbutamol along with her regular diabetes and thyroid medicines. She was initially managed in HDU, needed high flow oxygen support and strict fluid balance. In the ward, she suddenly became unresponsive and collapsed with an asystolic cardiac arrest. She developed severe metabolic acidosis with very high anion gap. The anion gap suggested the presence of a fixed acid and was not due to ketone production related to diabetes. This was confirmed at post-mortem also. She received prolonged resuscitation with multiple doses of adrenaline and boluses of bicarbonate. But remained in asystole and sadly succumbed. The pathologist noted that the child had a high anion gap metabolic acidosis and subsequent post mortem organic acid profile suggested that it was due to pyroglutamic aciduria. Pyroglutamic acidosis is noted to be a rare phenomenon which can happen in acutely unwell patients who receive antibiotics and paracetamol. The child had received paracetamol at a reduced dose in view of her renal impairment.

Conclusions The learning point from the above presentation is that Pyroglutamic acidosis (PGA) should be considered as a differential diagnosis in patients presenting with high anion gap metabolic acidosis with complex co-morbidities and risk factors. The mainstay of treatment of PGA includes cessation of medications that can influence the γ -glutamyl cycle or glutathione levels, N-Acetyl Cysteine (NAC) and the implementation of supportive measures. Unrecognised acidosis due to PGA can contribute to morbidity and mortality in patients with hepatic and renal dysfunction.