Methods The Illumina Trustight One Sequencing Panel was used for sequencing over 4800 genes known to be associated with a clinical phenotype and spanning 12 Mb of genomic content. The panel studied contains 125,000 probes based on the NCBI37/hg19 human reference genome. X-ray and MRI imaging were performed for fibular hemimelia.

Results The patient was born by elective cesarean section at 38 weeks with 2805 grams. He was the fourteenth pregnancy and ninth living baby of the 32-year-old mother. The infant's birth length was 45 cm, and the head circumference at birth was 35 cm. Physical examination of the patient revealed medial angulation in the right lower extremity from the knee, clubfoot deformity in the foot, and polydactyly in the left foot. There was no associated facial dysmorphism nor other associated anomalies apart from polydactyly. Abdominal, hip, and transfontanel USG and echocardiography were normal. Fibular hemimelia was found in the patient on x-ray and MR imaging. PMM2 and MEFV gene mutations were found in the gene analysis.

The patient was consulted to the orthopedic unit. Although limb amputation was recommended by surgeons, we investigated possible alternatives. As a result, the patient was referred to an external center for a tibial lengthening procedure.

Conclusions Congenital disorders of glycosylation are a group of hereditary diseases and they may present with different extremity anomalies.

Paediatricians with Expertise in Cardiology Special Interest Group

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EFFECT OF SERUM CHLORIDE AND SERUM SODIUM DERANGEMENTS ON DIURETIC RESPONSE IN CHILDREN ON CHRONIC FUROSEMIDE THERAPY

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Background Electrolyte disturbances are not uncommon in patients on chronic Furosemide therapy. The individual effect of both hyponatremia and hypochloremia has been studied in adults but not in the children with congenital heart disease and chronic congestive state.

Objectives We aimed at studying the occurrence of serum Chloride (Cl) and Serum Sodium (Na) abnormalities in children on prolonged Furosemide therapy and the possible relation of these abnormalities to the diuretic response in this population.

Methods The study included forty-five children, with congenital left to right shunts causing chronic congestive state which necessitated chronic Furosemide therapy. Patients in need to an increase in their Furosemide dose were recruited in the study. We assessed serum Cl and serum Na as well as parameters of diuretic responsiveness; net fluid output and change in body weight/40 mg Furosemide, and change in urinary Na/K ratio. These parameters were assessed initially and at day 3 after increasing Furosemide dose.

Results Median age of patients was 0.75 years, with 27 (60%) male patients and 8 (40%) female patients. According to serum levels of Cl and Na, patients were divided into four groups; isolated hyponatremia (15 patients, 33.3%), isolated

hypochloremia (9 patients, 20%), combined hypochloremia and hyponatremia (12 patients, 26.7%), and normal serum electrolytes (9 patients, 20%). Patients with combined hyponatremia and hypochloremia and those with isolated hypochloremia showed minimal clinical and radiological signs of decongestion as well as lowest changes in urinary Na/K ratio, fluid output and weight change/40 mg Furosemide on augmenting the diuretic dose, unlike the hyponatremic patients who had near normal parameters with no evidence of diuretic resistance.

Conclusions Both hypochloremia and hyponatremia are common in patients on prolonged Furosemide therapy. Hypochloremia is associated with a poor diuretic response, unlike isolated hyponatremia which does not seem to affect the diuretic response. Concomitant occurrence of hyponatremia and hypochloremia is associated with poor diuretic response as well which can be worse than that seen in isolated hypochloremia.

British Society for Rheumatology

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PROSPECTIVE UK AND IRELAND POPULATION-BASED STUDY OF JUVENILE-ONSET SYSTEMIC LUPUS ERYTHEMATOSUS

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Background Juvenile-onset systemic lupus erythematosus (JSLE) is a rare, multi-system autoimmune disease. SLE most frequently affects adults but onset is in childhood in up to 20% of cases. Where onset is in childhood, disease can be more severe, leading to significant associated morbidity and damage. **Objectives** This British Paediatric Surveillance Unit (BPSU) study of JSLE aims to describe presenting features, classification criteria, initial management and disease damage in the UK and Ireland population.

Methods BPSU methodology was used to identify new diagnoses of JSLE in children <18 years of age between September 2017 and September 2019. Relevant adult clinicians (including adolescent and adult rheumatologists, dermatologists and nephrologists) were also asked to report cases using parallel methodology. Data was collected at diagnosis and at one year follow-up. Data was analysed descriptively. McNemar's test was performed to compare sensitivities of different classification criteria.

Results 253 cases were reported with 131 incident cases included to date (122 exclusions: 60 duplicates; 26 diagnosed outside study period; 15 case definition not met; 19 no clinical information; 2 diagnosis removed at one year). Systemic Lupus International Collaborating Clinics (SLICC-2012) classification criteria were more sensitive for classifying patients with JSLE than American College of Rheumatology (ACR-1997) classification criteria (86.3% vs 75.6%, $p < 0.05$). European League Against Rheumatism/ACR (EULAR/ACR)-2019

classification criteria demonstrated similar sensitivity to SLICC-2012 (84.4%, $p=0.29$) and were superior to ACR-1997 ($p<0.05$). However, 10.9% of patients met no classification criteria at diagnosis.

The following results describe the 87.0% of patients meeting at least one of ACR-1997 and/or SLICC-2012 at diagnosis. The median age at diagnosis was 13.9 years. The majority of patients were female (7:1), and this ratio was maintained in the youngest patients. Non-Caucasian children were over-represented (63%). Median time to diagnosis was 4.0 months but wide variation was seen with diagnosis taking over 12 months in 21.1%. Most patients were diagnosed by rheumatology (66.1%) but a significant proportion by general paediatricians (15.2%). Presenting features were consistent with previous literature. Multisystem involvement was assessed using the domains described in the paediatric adaptation of the British Isles Lupus Assessment Group index. Multisystem involvement was common as expected (median number of systems involved was 4). Fatigue affected most patients (84.2%). 23.3% of patients had damage on the SLICC damage score at diagnosis and 12.5% of patients had acquired damage within the first year following diagnosis. All patients received systemic immunomodulatory treatment. The majority of patients (88.6%) received systemic glucocorticoids at diagnosis, with 69.9% of patients continuing to receive glucocorticoids at one year.

Conclusions Challenges in classifying patients with JSLE remain but this study supports the use of SLICC-2012 classification criteria as the best available system. The significant burden of JSLE is clearly exemplified by the high number of patients accruing disease damage within one year of diagnosis, and the significant ongoing glucocorticoid use. Further work will describe the minimum incidence of JSLE and diagnostic delay.

British Society of Paediatric Gastroenterology, Hepatology and Nutrition

809 AUDIT OF THE THERAPEUTIC DRUG MONITORING OF INFLIXIMAB IN PAEDIATRIC INFLAMMATORY BOWEL DISEASE

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Background Biologics have emerged as new treatment options for IBD, targeting the immune system and its products. This has revolutionised the way that IBD is managed. It is important that serum drug levels are monitored regularly to ensure the biologics are administered at therapeutic doses. If levels are not regularly tested, they may be sub-therapeutic. This increases the risk of a flare up or development of anti-infliximab antibodies.

Objectives Guidance for therapeutic drug monitoring varies between different centres. Local guidance indicates monitoring should be done pre 3rd or 4th dose. The aim of the audit was to compare practice at the GNCH to the guidance.

Methods Information regarding the timing of therapeutic drug level sampling was analyzed retrospectively, using data from electronic records, for all patients receiving Infliximab at

GNCH. Levels were judged to be 'on time', according to trust guidance, if they were taken before the 3rd or 4th doses of infliximab administration. This process would reset once serum infliximab levels were taken. Trust guidance has been determined by specialist discussion between other sites that use and monitor infliximab in paediatric IBD. This has been influenced by advice from ESPGHAN and the PANT study.¹

Results 87 patients have started on Infliximab between 2012 and 2019, aged 4 to 17 years old. 5/87 started too recently to have 4 doses, thus were excluded. Overall, 34% of patients had all their levels taken accurately, with 53% of total levels taken accurately (table 1). The improvement in accuracy coincides with the introduction of a specialist pharmacist to oversee biologic application.

Additionally, patients who started infliximab prior to 2018 had poorer accuracy in their 2019 testing with accuracies of 36, 43 and 53% in patients started in 2015, 2016 and 2017 respectively. Compared to 65% accuracy in 2018 and 71% in 2019 (table 2).

To comment on 'early' levels, although this clinically does not lead to poorer care, according to the guidelines it is over testing and an unneeded use of resources.

Of all the levels taken, 56.2% showed serum level lower than was deemed therapeutic, thus showing how important regular testing is.

Abstract 809 Table 1

	Accuracy of levels taken between 2013–2017 (total 86)	Accuracy of levels taken between 2018–2020 (total)	Accuracy of all levels taken
On Time	35%	59%	53%
Early	34%	20%	23%
Late	31%	21%	24%

Conclusions While infliximab monitoring in GNCH has improved over the time it has been conducted there is still room for improvement. Part of this should come from further guidance from the specialist pharmacist.

My suggestions for making further improvements are:

- Increased awareness of the importance of infliximab monitoring in patients and healthcare providers.
- A messaging system, similar to the one used for Azathioprine, to remind patients and day case units when they require a blood test.
- Infliximab blood test forms should be made electronic, like other blood tests, in order to improve accessibility.

In further audits it may be informative to explore how these levels were used, compared to algorithms/recommendations set by studies like that conducted by Vaughn *et al.*²

REFERENCES

1. Kennedy NA, Heap GA, Green HD, *et al*; UK Inflammatory Bowel Disease Pharmacogenetics Study Group. Predictors of anti-TNF treatment failure in anti-TNF-naïve patients with active luminal Crohn's disease: a prospective, multicentre, cohort study. *Lancet Gastroenterol Hepatol* 2019;4:341–353.
2. Vaughn BP, Sandborn WJ, Cheifetz AS. Biologic concentration testing in inflammatory bowel disease. *Inflamm Bowel Dis* 2015;21:1435–42.