The changes in all parameters and discontinuation rates in the two groups were compared after one and four months of the therapy.

**Results** In total 71 patients were included in the study, 18 patients of them lots follow up during the study, while 53 continued. There were no significant differences in the baseline demographic and biochemical baseline data between the two groups (P > 0.05).

After one month, there was statistically significant difference between the two groups in the PGS as it decreased by <1.5x base line in the T-group compared to the C-group (P < 0.02).

After four months, there were statistically significant between the 2 groups regarding decreased ALT levels below 1.5x base line levels, AST, GGT and bile acid levels in favour of the T-group (P < 0.02, 0.047, 0.026 and 0.001 respectively).

**Summary and Conclusion** The use of FF in combination with UDCA provided satisfactory clinical outcomes, which could be a promising alternative, but patients should be monitored closely as side effects may occur despite achieving improvements in pruritus.

**REFERENCES**

**P23**

**FIRST REPORTED CASE OF AN INTERLEUKIN-2 RECEPTOR ß DEFICIENCY IN AN INFANT BORN TO NON-CONSANGUINEOUS PARENTS, PRESENTING WITH FAILURE TO THRIVE AND ENTEROPATHY**

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**Introduction** Failure to thrive is not an uncommon paediatric presentation with multiple, often easily treatable, aetiologies. Immune dysregulation is a rare cause which often manifests with a combination of inflammatory and infectious pathology. Aim We present a rare cause of immune deficiency, the first reported case attributable to this genetic abnormality in the absence of consanguinity, presenting with immune mediated enteropathy.

**Method** A five-month-old female presenting with bronchiolitis symptoms, was found to be failing to thrive. Her weight was on the 0.4th centile, gaining just 200 g in the preceding eight weeks.

The first born to non-consanguineous parents of Pakistani descent, she was born at 39 weeks’ gestation weighing 2.52 kg. There was no significant family history and she had breastfed successfully for the first three months, growing along the 9th centile, and meeting developmental milestones.

By seven months she was 1 kg below the 0.4th centile, weighing the same as at three months. Bottle refusal and a failure to wean to solids accompanied chronic diarrhoea, oral and perianal inflammation, and eczematous dermatitis. Amino acid nasogastric feeds resulted in a small weight gain, but no relief of systemic symptoms.

Developmental delay and soft dysmorphic features were apparent, and she developed recurrent fevers, polyarticular swelling affecting knees and joints of the hands and feet, widespread lymphadenopathy, and chronic bilateral uveitis.

**Results** Double negative T cell levels were elevated, inflammatory markers and liver enzymes were persistently high and cytomegalovirus (CMV) IgG was positive. Hypergamma globulinaemia and numerous autoantibodies were demonstrated. Fae calprotectin was markedly raised, and endoscopy revealed nonspecific acute on chronic inflammation of the duodenum and stomach.

By 12 months of age, total parenteral nutrition and pulsed methylprednisolone, followed by oral prednisolone had been started. Symptoms improved, she began to gain weight and make developmental progress.

Full genome sequencing and parental genetic analysis, identified an autosomal recessive interleukin-2 receptor ß (IL2RB) mutation, giving a diagnosis of IL2RB deficiency.

The patient is now recovering following haematopoietic stem cell transplantation, with the aim of definitive cure.

**Conclusion** Interleukin-2 is involved in building protection against autoimmune disease by stimulating T-cell differentiation. Genetic mutations in its receptor are exceptionally rare, with only four homozygous defects identified in seven infants, one neonate, and two fetuses, all conceived to consanguineous marriage partners. Common clinical manifestations amongst the seven surviving the neonatal period reflect the underlying immune dysregulation with enteropathy, arthritis, uveitis, dermatitis and hypergamma globulinaemia, together with a susceptibility to respiratory and herpesvirus infections. Inflammatory features were prominent in this case but she went on to develop symptoms of CMV illustrating the conundrum of managing inflammation with underlying immunosuppression.

Failure to thrive, diarrhoea, eczema and recurrent viral infections are not uncommon in paediatrics. However, when rare aetiology is responsible, the journey from presentation to diagnosis can be long.

The time from presentation to referral for definitive treatment was remarkably quick in this case. This is testament to the exceptional collaborative multidisciplinary working across sub specialties, leading to a prompt diagnosis and definitive management.