**Abstract G63 Table 1**

| Grade of Reflux | Number of cases n = 39 |
|---|---|
| 1 | 2 (5%) |
| 2 | 25 (64%) |
| 3 | 8 (21%) |
| 4 | 4 (10%) |

**Abstract G63 Table 2**

| VUR | Number of cases n = 39 |
|---|---|
| Bilateral | 26 (67%) |
| Right | 6 (15%) |
| Left | 7 (18%) |

We had discharged 5 children (13%); in 4 of these VUR has been demonstrated to have resolved; and 34 (87%) are currently being followed up. Of these, 2 have scarring with recurrent UTIs and 1 has a scarred kidney but VUR has resolved.

**Conclusion** 19% of asymptomatic infants screened for VUR because of a positive family history of VUR or RN have themselves got VUR, with the majority (67%) having bilateral VUR. By identifying these cases resources and education are targeted to those families to encourage rapid diagnosis and treatment of UTIs with the ultimate aim of preventing scarring.

## G64(P) CO-CREATING A CO-ORDINATED COMPLEX CARE PLAN TO IMPLEMENT IN A PAEDIATRIC RENAL SERVICE

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**Aim** NHS Kidney Care commissioned project to develop and implement a patient/family held care plan to be created by service users and multi-agency staff.

This project aimed to review existing documentation with staff and service users and co-design a care plan, to improve co-ordination of care across agencies and standardise access to education and information resources. Promotion of self-management and partnership working with parents, carers and young people was central to this.

**Methods** A Review, Agree, Implement and Demonstrate (RAID) model was used to develop and trial the care plans. Mixed methods were used to obtain qualitative data in the review and agree stages. Professionals and service users were invited to engage via questionnaires, interviews and co-creation events.

**Results** articipation data

**Abstract G64 Table 1**

| Method | Families: Number offered | Families: Number participating | Professionals: Number offered | Professionals: Number participating |
|---|---|---|---|---|
| Questionnaire | 76 | 34 (44%) | 84 | 32 (46%) |
| Co-creation event and interviews | 34 | 10 (29%) | 32 | 13(41%) |

Questionnaire results indicated that 58% of families are using some form of hand held record created for themselves.

A draught care plan was co-created by attendees at the focus group and interviewees. They then undertook a review and an amended care plan was collaboratively developed. The resulting care plan is transportable for use in a variety of settings. It is designed to encourage the patient and family to be active participants in care planning, and suitable for use from childhood through to young adulthood. The trial of the draught care plan is in progress with 20 patients and the multi-agency teams working with these children/young people. The utility and value of the care plan will be assessed after a minimum of 3 months use. A further review will be undertaken prior to full implementation

**Conclusion** This project has demonstrated the willingness of families and professionals from health, social and education services to participate in a co-creation project. This has allowed us to develop draught documentation designed to support the health, education and social development of children with complex renal disease.

## G65(P) UNILATERAL HYPOPLASTIC KIDNEY – A NOVEL AND HIGHLY PENETRANT FEATURE OF FAMILIAL JUVENILE HYPERURICAEMIC NEPHROPATHY

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**Aim** To highlight an interesting and novel renal phenotype that may provide an insight into the genetics surrounding the development of isolated renal hypoplasia.

**Methods** A sixteen year old boy was referred to Paediatric Nephrology services following concerns of a strong family history of renal disease. Both his mother and maternal aunt have end stage renal disease. Two of his maternal cousins were found to have chronic kidney disease. All affected members had evidence of hyperuricaemia. The patient's grandparents and maternal uncles were not affected. Renal ultrasounds performed on affected family members revealed unilateral renal hypoplasia in the index case, as well as his mother and aunt.

**Results** Our case report describes a pedigree with familial juvenile hyperuricaemic nephropathy, a relatively uncommon condition characterised by hypoexcretion of urate leading to hyperuricaemia, gout and progressive renal impairment. This family, however, require our attention for several reasons. Firstly, three affected family members demonstrate unilateral renal hypoplasia inherited in an autosomal dominant manner: to our knowledge this is the first report to describe such a phenotype. Secondly, two affected cousins had normal sized kidneys, suggesting a modifier gene effect, and lastly affected members have tested negative for mutations in two of the major genes implicated in FJHN, which have also been linked to a role in renal morphogenesis: uromodulin (*UMOD*) and hepatocyte nuclear factor 1β (*HNF1β*).

**Conclusion** Isolated renal hypoplasia is a common congenital anomaly for which a gene association has never been found. The presence of this phenotype in an autosomal dominant manner in this pedigree is therefore of great potential importance, for the ability to identify for the first time a gene responsible for unilateral renal hypoplasia. The association here with renal failure and hyperuricaemia is fascinating, and may provide novel developmental insights linking tubular development, control of lateral renal maturation and renal size. We discuss the known genetics surrounding renal embryogenesis and the implications our pedigree may have for further understanding of this common developmental anomaly.

## G66(P) TO DETERMINE THE ACCURACY OF PHASE CONTRAST MICROSCOPY IN PREDICTING URINE CULTURE RESULTS

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**Aims** To determine the accuracy of phase contrast microscopy in predicting urine culture results.

**Design and methods** A prospective study comparing the results of phase contrast microscopy interpretation of urine samples with that of urine culture. Samples were microscoped at the time culture was being performed.

The sample size though ongoing was based on the availability of the clinician to get to the laboratory at the time of urine culture. Uncentrifuged, unstained urine samples were examined with an Olympus BH2 microscope enabled with phase contrast at 400x magnification mounted on a Hawksley Nebauler counting chamber. Samples were interpreted as being either "Positive," "Negative" or "Indeterminate" based on their level of bacteriurea. "Indeterminate" specimens would be repeated in the clinical setting and were not further analysed in our laboratory based study.

**Results** 65 samples were microscoped. Immediate determination was made for 62 samples (95.4%). 3 samples were deemed as "Indeterminate."

**Abstract G66 Table 1**

| | Culture Positive | Culture Negative | Total |
|---|---|---|---|
| Microscopy Positive | 15 | 3 | 18 |
| Microscopy Negative | 2 | 42 | 44 |
| Total | 17 | 45 | 62 |

Of the 62 microscopy interpretations 57 (91.9%) showed concordance with the microbiology culture results. Sensitivity of 15/17 (88.2%), specificity 42/45 (93.3%), positive predictive value 15/18 (83.3%) and negative predictive values of 42/44 (95.5%) were obtained. Video evidence is available showing bacteria in 2 of the samples deemed "Microscopy Positive" but "Culture Negative." If these samples, as they ought to, returned as "Culture Positive" then microscopy would have attained concordance 59 (95.2%), sensitivity 17/19 (89.5%), specificity 42/43 (97.7%), positive predictive value 17/18 (94.4%), negative predictive value 42/44 (95.5%).

**Conclusions** Phase contrast microscopy afforded immediate interpretation in 62/65 (95.4%) of samples studied. Microscopy interpretation showed high concordance rate, sensitivity, specificity, positive and negative predictive values when compared to the accepted gold standard i.e., microbiological culture even with the apparent error in the culture process.

## G67(P) DENT'S DISEASE COMPLICATED BY AN ACUTE BUDD-CHIARI SYNDROME: CASE REPORT

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We present the case of a young boy with Dent's disease, identified as having a mutation in the kidney-specific chloride-proton antiporter CLCN5 during investigation for nephrotic range proteinuria. He went on to develop a growth hormone deficiency requiring treatment with recombinant growth hormone followed by an acute presentation with hepato-renal failure and thrombotic occlusion of both middle and right hepatic veins consistent with a diagnosis of Budd Chiari syndrome, which required a prolonged period of intensive care. We have identified 3 reports in the literature in which growth hormone therapy has been used to treat short stature associated with Dent's disease. This report confirms that growth hormone deficiency is a recognised finding in this rare disease. There are no previous reports on thrombotic complications associated with either Dent's disease or the use of recombinant growth hormone *per se*. The cause of the Budd-Chiari syndrome in this case has yet to be fully elucidated, but potentially widens the Dent's phenotype and gives support to the previous observation that this disease is multifaceted with a possible role for as yet unidentified environmental and/or genetic modifying factors.

## G68(P) VESICOURETERIC REFLUX – AN UNUSUAL GENETIC ASSOCIATION OF COMMON CONDITION

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**Background** Vesicoureteric reflux is the most common urological anomaly in children and is a common cause of end stage renal failure and hypertension in children. It usually occurs in isolation and in vast majority of cases it improves on its own. However rarely it can be associated with coloboma and renal hypoplasia, consequent to PAX2 gene mutation.

**Case Report** We report a case of 14 year old boy, who was under regular paediatric follow-up since the age of 5 month when he had first episode of urinary tract infection. He was put on trimethoprim prophylaxis and micturating cystourethrogram (MCUG) revealed bilateral grade 3 reflux without obstruction at the age of 8 months. He was lost to follow up for 3 years, delaying investigation. Dimercaptosuccinic acid (DMSA) scan at 4 years of age showed small right kidney with no scarring and differential function of 33%. On routine eye check at age of 5 years he was noted to have bilateral optic disc coloboma, however his vision was not affected. He was noted to have mild impairment in renal function at age of 6 yrs with eGFR of 63ml/min/1.73m$^2$ which remained stable for next 6 year. During this period he also became significantly obese (body mass index 33) with significant disproportion. He remained normotensive during this period. His renal function started deteriorating for past couple of years with the development of proteinuria. His diagnosis was revisited and was thought to be due to PAX2 gene mutation.

**Discussion** Paired box (*PAX*) genes play a critical role in human development and disease. The *PAX2* gene is expressed in primitive cells of the kidney, ureter, eye, ear and central nervous system and is required for normal kidney and eye development. Mutation in PAX2 gene has been described in the several families with optic nerve coloboma, renal hypoplasia, mild proteinuria and vesicoureteric reflux.

**Conclusion** We report this case to raise awareness amongst paediatricians about this uncommon genetic association of vesicoureteric reflux with renal hypoplasia and coloboma which can lead to progressive renal failure.

## G69(P) PAEDIATRIC RENAL TRANSPLANTATION WITH BARDET-BIEDL SYNDROME (BBS) AND SITUS INVERSUS TOTALIS

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**Introduction** Bardet-Biedl syndrome (BBS) is a rare autosomal recessive disorder characterised by a genetic dysfunction that causes cystic malformation of the kidneys alongside features such as postaxial polydactyly, central obesity and a spectrum of learning difficulties. A rare cause of renal failure in children that ultimately requires transplantation at a very young age. We report the first case of successful renal transplantation in a 3 and a half year old child with both BBS and situs inversus totalis .