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

doi:10.1136/archdischild-2013-304107.076

V Bates, J Woodland, S Dolby, C Inward. *Renal Unit, University Hospital of Bristol, Bristol, UK*

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 participation 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

doi:10.1136/archdischild-2013-304107.077

LA Plumb, M Marlais, MA Saleem. *Department of Paediatric Nephrology, University Hospitals Bristol NHS Foundation Trust, Bristol, UK*

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 hypoxcretion 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 1 β (*HNF1 β*).

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

doi:10.1136/archdischild-2013-304107.078

¹VRS Singh, ²J Ramesar, ³S Maharaj, ³S Ramsewak, ⁴J Dean, ⁵C Cave, ⁶S Mayers. ¹Child Health Unit, University of the West Indies, St Augustine, Trinidad and Tobago; ²Paediatrics, Eric Williams' Medical Sciences Complex, Champ Fleur, Trinidad and Tobago; ³University of Sheffield, Sheffield, UK; ⁴University of East Anglia Medical School, Norwich, UK; ⁵University of Oxford, Keble College, Oxford, UK; ⁶Microbiology, Eric Williams Medical Sciences Complex, Champ Fleur, Trinidad and Tobago

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 bacteriuria. "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

doi:10.1136/archdischild-2013-304107.079

¹C Platt, ²J Dudley, ³J Hartley. ¹General Paediatrics, Gloucestershire Hospitals NHS Foundation Trust, Gloucester, UK; ²Paediatric Nephrology, United Hospitals Bristol NHS Foundation Trust, Bristol, UK; ³Department of Hepatology, Birmingham Children's Hospital, Birmingham, UK

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

doi:10.1136/archdischild-2013-304107.080

M Gupta, R Wasala. Department of Paediatrics, Sandwell and West Birmingham Hospitals NHS, West Midlands, UK

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/1.73m² 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

doi:10.1136/archdischild-2013-304107.081

¹JH Frost, ²A Bagul, ³IH Mohamed, ³MG Mould, ⁴HK Jahn, ⁵G Koffman. ¹Oxford University Hospitals NHS Trust, Oxford, UK; ²Guys and St Thomas NHS Foundation Trust, London, UK; ³Royal North Shore Hospital, Sydney, Australia; ⁴Leicester Royal Infirmary, Leicester, UK

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 post-axial 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.