# Oral / Poster Presentation

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## First day Oral Presentations

### Tues- O1

### Genetic Aspects of Distal Renal Tubular Acidosis (dRTA) Update

Sharifian M, Esfandiar N, Mazaheri S, Kariminejad A, Mohkam M, Dalirani R

Shaheed Beheshti University of Medical Sciences, Tehran, IRAN.

**Introduction:** Distal renal tubular acidosis (dRTA) is characterized by impaired renal H+ secretion resulting in metabolic acidosis. In Autosomal recessive dRTA, a substantial fraction of patients have progressive bilateral sensorineural hearing loss. This coexistence is due to the mutations of a gene expressed both in the intercalated cells of the kidney and the apical membrane of the interdental cells and the epithelium of endolymphatic sac in cochlea. The gene ATP6V1B1, is responsible for encoding β subunit of the $H^+$-ATPase pump which plays an important role in the maintenance of acid- base homeostasis and endolymphatic acid secretion.
The aim of this study was to assess the mutations in ATP6V1B1 gene in our patients with dRTA.
**Materials & Methods:** In this study 52 children admitted with the diagnosis of RTA at nephrology clinic were evaluated. Diagnosis of dRTA was based on clinical manifestations and detection of normal anion gap metabolic acidosis, hypokalemia and urine PH which was never under 5.5 and positive urinary anion gap. Audiometry was performed in all patients with dRTA who were cooperative and evoked potential in infants for hearing evaluation. Sequencing of the ATP6V1B1 gene was performed in patients with dRTA and sensorineural hearing loss. Statistical analysis was performed using Excel software by Student t test and differences with P value lower than 0.05 were considered significant.
**Results:** Twenty eight patients (53.8%) had dRTA. Fifty one percent were under the age of one and 49% were between 1-12 years. Sixteen patients (57%) were male and 12 (43%) female. Twelve patients (42%) had bilateral sensorineural hearing loss consisting 6 of 15 boys (40%) and 6 of 12 girls (50%). There was no correlation between hearing loss and the gender (P= 0.38). Four patients with hearing loss had mutation in ATP6V1B1 gene (14% of patients with dRTA and 33% of patients with dRTA and hearing loss).
**Conclusions:** This study indicated that significant percentage of the children with dRTA had sensorineural hearing loss and mutation in ATP6V1B1 gene. It is recommended to perform audiometry in all children with dRTA.

### Tues - O2

### Causes of Fanconi Syndrome in Children's Hospital of Tabriz

Mortazavi F, Ghalegolab-Behbahan A

Tabriz University of Medical Sciences, Tabriz, Iran

**Introduction:** Fanconi syndrome (FS) is a generalized dysfunction of renal proximal tubules leading to urinary wasting of glucose, amino acids, bicarbonate, phosphate and other solutes. The causes of FS are divided into: hereditary, acquired and exogenous substances. The aim of this study was to detect the causes of FS in children admitted in Children's Hospital of Tabriz.
**Materials & Methods:** In a descriptive and cross sectional study medical documents of all children who admitted in Children's Hospital of Tabriz from 2000-2010 with diagnosis of FS were studied. Demographic characteristics and clinical and laboratory findings were gathered in designed form.
**Results:** During ten years 39 patients with FS were diagnosed. Age of patients at the diagnosis was 2 months- 9 years (mean: 23.6± 12.7 months). Twenty three patients were male (58.9%) and 16

patients (41%) were female. Consanguinity was present in 23 (64%) cases. The causes of FS in a descending order were: cystinosis in 14 patients (36%) , tyrosinemia in 5 (12.8%), glycogen storage disease type I (Von-Gierke) in 4 (10%), galactosemia in 3 (7.6%), chemotherapy with cisplatin in 3 (7.6%), Fanconi-Bickel syndrome in 2 (5%), idiopathic FS in 2 (5%), nephritic syndrome in 2 (5%), Wilson disease in 1 (2.5%), Dent disease in 1 (2.5%), intoxication with heavy metals (mercury) in 1 (2.5%), and side effect of Na-valproate in 1 (2.5%) patient. Failure to thrive was observed in all patients with hereditary FS and in 89.7% of all patients. Nine patients (23%) progressed to end stage renal failure and 7 patients (17.9%) died.

**Conclusions:** In our area hereditary FS is more common than acquired forms and among them cystinosis is the most common cause with a poor prognosis.

**Tues -O3**

## Plasma Neutrophil Gelatinase-associatde Lipocalin (NGAL) in Infants With and Without Sepsis Who Admitted in Ali-Asghar Hospital during 2011

Khosravi N, Karimi H,

Iran University of Medical Science, Tehran, Iran

**Introduction:** Neutrophil gelatinase-associated lipocalin (NGAL) is an inflammatory maker of cell injury which increases significantly in situations such as ischemic damage, cytokine release, and sepsis. There are different studies about the relationship between serum levels and urinary levels of NGAL and acute kidney injury (AKI) and sepsis in adult and pediatric population. The aim of this study is to evaluate plasma NGAL levels in the diagnosis of neonatal sepsis.

**Materials & Methods:** In this study, 120 neonates who admitted in NICU of Ali-Asghar hospital included in the study. They were divided in two group; neonates with sepsis (n=52) and control group (n=68). Serum levels of NGAL in the two groups (cases/controls) were evaluated on admission, using (NGAL Rapid ELIZA Kit kit 037) and were measured by ELISA. NGAL levels in both groups were compared. Analysis for the ROC cut off level of serum NGAL in diagnosis neonatal sepsis and its diagnostic accuracy was calculated.

Results: 120 infants were examined in this study (56.7% male) and mean gestational age of them was 36.7±3.7 weeks (range 26 to 40 weeks). Mean NGAL plasma level was significantly higher in patients with sepsis than controls (102.9±65.9 vs. 29.2±13.3 ng/ml; P= 0.0001). Plasma NGAL was significantly higher in male infants than female infants (respectively, 73.5±68.7 vs. 45.1±32.3 ng/ml; P= 0.007) and also it has positive relationship with the duration of hospital stay (r=0.640, P=0.0001). In the cut-off point of 48 ng/ml, plasma NGAL had 92% sensitivity, 91% specificity, and accuracy of 80%, which it seems to be the most appropriate cut-off point of plasma NGAL for diagnosis of neonatal sepsis.

**Conclusions:** Regarding to high sensitivity, specificity and accuracy of NGAL plasma levels, it seems a good biomarker for early diagnosis of neonatal sepsis.

**Tues -O4**

## Assessment of Prognostic Factors in Children RTA Type IV Overwhelmed By Bilateral Obstructive Uropathy.

Yousefichaijan P, Firouzifar M,

Arak university of Medical sciences, Arak, Iran

**Introduction:** Hyperkalemic (type IV) renal tubular acidosis occurs secondary to impaired renal responsiveness to Aldosterone. This can occur chronically in infants and children with a history of obstructive uropathy. The aim of this study is to assess contributory factors in improving conditions like hyperkalemia and metabolic acidosis as well as growth status in children suffering from bilateral obstructive uropathy and RTA type IV.

**Materials & Methods:** In this study, we recruited and observed 48 patients affected with both bilateral obstructive uropathy at urinary bladder outlet and RTA type IV for two years, during, children's growth, sonographic data, renal function and serum electrolytes underwent serial assessment and in case of clinical indication, the patients were treated with drugs like citrate sodium and Kayexalate. Noteworthy patient's death resulted in exclusion from the study.

Results: Frequent urinary tract infection (p=0.0011), infants and children with abnormal <20 weeks gestational sonography results like bilateral hydronephrosis (p=0.00001), birth weight below 2500 gr (LBW) (p=0.0014), preterm delivery (p=0.001), maternal age below 20 years (p=0.0018), pregnancy more than two times (p=0.004), admission due to respiratory problems during infancy period (p=0.003) were discretely accompanied with no disease convalescence while other factors were associated with no significant difference.

**Conclusions:** Regarding the paper results, it seems logic to consider abortion in case of renal hydronephrosis and dysplasia in gestational age below 20 weeks. Moreover, medical care during pregnancy for a term delivery with suitable weight (resulting in better maturation of lung parenchyma and precluding hypoxia related renal injuries) prevents

**Tues -O5**

## Distal Renal Tubular Acidosis: From Clinics to Molecular Mechanisms

Mohebbi N[1], Wagner CA[2]
[1]Division of Nephrology, University Hospital Zurich.

[2]Institute of Physiology, University of Zurich, Zurich, Switzerland.

Systemic acid-base homeostasis is the product of complex interactions between metabolism, regulated exhalation of $CO_2$ by the lungs, and acid or base excretion by the kidneys. The importance of renal acid-base transport has been highlighted by mutations identified in several proteins involved in this task in patients with inherited forms of renal tubular acidosis. In humans, distal renal tubular acidosis (dRTA) type I can be caused by mutations in the anion exchanger AE1 (SLC4A1) and the B1 and a4 subunits of the V-type $H^+$-ATPase. Type I dRTA is usually characterized by hypokalemic hyperchloremic metabolic acidosis. To better understand the pathophysiology of dRTA and the underlying mechanisms of defective distal urinary acidification we have generated two different mouse models that are genetically altered for the *AE1* and *B1* subunit genes. Furthermore, we developed several rat models to study mechanisms contributing to acquired forms of dRTA.

**Tues -O6**

## Evaluation of Effect of Heparin Sodium a Diluting Factor on Blood Gas Analysis Outcomes

Pournasiri Z, Hatami H

Shahid-Beheshti University of Medical Science, Tehran, Iran

**Introduction:** Sodium heparin solution is widely used as an anticoagulant in blood gas analysis. Although alterations in blood gas measurements may occur when small sample volumes are diluted by heparin. So we aimed to determine correlation between heparin diluted status and Blood gas analyses.

**Materials & Methods:** A cross sectional study was conducted on 100 pediatric ward admitted patients. We take two different samples by similar syringes, one contained 5% extra heparin sodium. Both blood samples were analyzed and compared together.

**Results:** Our results shown that heparin contained samples had wrong blood gas analyses reports which acidotic axis may had shown imbalance cause of $H^+$ imbalance.

**Tues -O7**

## Fanconi-Bickel Syndrome Versus Osteogenesis Imperfecta: An Iranian Case With A Novel Mutation In Glucose Transporter 2 Gene, And Review Of Literature

Hadipour F [1], Sarkheil P[1], Noruzinia M[2], Hadipour Z[1], Baghdadi T[2,4], Shafeghati Y[1,3]

[1]Sarem Cell Research Center , [2] Tarbiat Modares University,[3] University of Social Welfare Science and Rehabilitation,[4]TMSU,Tehran, Iran

**Case report:** Fanconi-Bickel syndrome is an extremely rare hereditary metabolic disease, characterized by hepatomegaly due to glycogen storage, refractory hypophosphatemic rickets, marked growth retardation and proximal renal

tubular acidosis. Recurrent bone fractures are one of the hallmark findings. It is a single gene disorder; the responsible gene belongs to the facilitative glucose transporters 2 (GLUT2) family gene or (SLC2A2) mapped to the q26.1-26.3 locus on chromosome 3, and encodes the GLUT protein 2. This protein is expressed in pancreatic ί-cells, hepatocytes, renal tubules, and intestinal mucosa. Several mutations in the GLUT2 gene have been reported in different ethnicities. Herein we report an Iranian girl with a missed diagnosis of osteogenesis imperfecta. She was referred with the history of frequent fractures, and severe motor delay and was suspected to osteogenesis imperfecta. Following the case we detected refractory rickets instead of OI, sever growth failure, proximal renal tubulopathy and RTA, and enlarged kidneys, progressive hepatomegaly, and GSD on liver biopsy. Glucose and galactose tolerance tests confirmed abnormal carbohydrate metabolism. Molecular analysis on GLUT2 gene revealed a homozygous novel mutation in exon 5; it was 15 nucleotide deletion and 7 nucleotide insertion and caused a frame shift mutation, produced a premature truncated protein (P.A229QFsX19). This mutation has not been reported before in the relevant literature.

**Tues - O9**

## Phenotype and Outcome of Bartter Syndrome in Iranian Children: Single Center Experience.

Hooman N, Otukesh H, Razzaghy-Azar M, Hoseini R, Nickavar A.

Iran University of Medical Sciences, Tehran, Iran

**Introduction:** Bartter syndrome is a rare inherited tubulopathy that is characterized by renal salt wasting, hypokalemic metabolic alkalosis, normal blood pressure despite of high blood level of renin and aldosterone. Here, we describe the phenotype- based classification of children diagnosed as bartter syndrome and the outcome of these cases.

**Materials & Methods:** This is a retrospective study. The data of all of the patients recorded as ICD E268, admitted in pediatric nephrology department between 2004 and 2013, were reviewed. The inclusion criteria were: 1- hypochloremic metabolic alkalosis, 2- normal blood pressure, 3- high urine chloride (>20 mEq/L), 4- high renin and aldosterone. Demographic data, laboratory tests, and the outcome were recorded.

**Results:** From 23 children who had ICD E268 code, 19 children (10 males,9 females) fulfilled the criteria. The mean (range) age of presentation was 3.2 m (birth-12 m), the mean birth weight was 2.7 kg (1.5-4.5 kg), and the mean gestational age was 33.14 wks (28-38 wks). The most common manifestations were prematurity (83.3%), polyhydraminios (78%), growth retardation and failure to thrive (94.7%), delayed cognitive development (31.6%), polyuria/ polydipsia or dehydration episodes (94%), muscle weakness or cramps (17.6%), nephroclacinosis (22%), sensorineural deafness (16.7%), temporarily high blood pressure in one. Consanguinity was reported in 58% and affected siblings were in 39%. The classifications of patients according to their phenotype were: aBS-I-II (58%), cBS-III (21%), SND-IV (16%), CaSR-V (5%). The patients were followed for mean 7.95 years (range: 9 m – 17 yrs). One (6%) died of recurrent sepsis, one (6%) fully recovered with no need to medication while his brother with the same diagnosis led to ESRD. Two (12%) cases were transplanted, and the others have glomerular filtration rate between 60-90 ml/min/1.73 $m^2$. The observed co-morbidities were hyperlipidemia and obesity in two, severe varicose in leg and tight and gynecomastia in one, tonic-colonic generalized seizure and spasticity in one.

**Conclusions:** Bartter syndrome is more prevalent due to consanguinity marriage in Iran. However it can be classified clinically, but genetic analysis is more powerful for early diagnosis and start proper treatment.

**Tues - O10**

## Relationship between Serum Level of Calcium and Urinary Levels of Calcium, Citrate and Urine Ph in Children with Renal Stones

Akhavan-Sepahi M,

Qom University of Medical Sciences, Qom, Iran,

**Introduction:** Prevalence of urolithiasis in childhood is increasing. Many children with stone disease have a metabolic abnormality. Hypercalciuria and hypocitraturia are considered

the most important risk factors for urolithiasis. The main aims of this study were to assess serum level of Ca, calciuria, citraturia and urinary pH and to determine whether urinary Ca concentration is a helpful biomarker in metabolic evaluation of children with renal stone.

**Materials & Methods:** This cohort study was performed at Qom University of Medical Sciences on 100 pediatric patients with documented urolithiasis. We collected 24-h urine samples from 100 stone-forming children and adolescents with hypocitraturia and from 121 healthy controls. Urinary calcium, pH, citrate, and oxalate were assessed and compared between the two groups. Data were analyzed using SPSS-13 software.

**Results:** Patient's age was ranged from 20 days to 14y/o with mean age of 3.32+2.53 years; 54 patients were male (54%) and 46 patients were female (46%).In both stone-formers and controls, hypercalciura was inversely related to citraturia and urinary pH. Metabolic disorder was detected in 95% of patients; the most prevalent urine metabolic abnormalities were hypocitraturia (56.8%), hypercalciuria (29.4%), hyperuricosuria (26.3%), hyperoxaluria (14.7%), phosphaturia (8.4%) and cystinuria (6.3%).

**Conclusions:** This study similar to other studies in Iran have shown that the prevalence of hypercalciuria is significantly higher compare to other countries, it may be associated with excessive intake of calcium and sodium. Compared to controls, stone-formers with hypocitraturia demonstrated a higher urinary Ca concentration, but this was proportional to calciuria. However, the Ca/Citrate ratio may be a useful clinical tool in evaluating children with urolithiasis.

**Tues -O11**

## Normal Ca/Cr, Na/Cr and K/Cr Ratio in Healthy Adolescents (North Of Iran)

Sorkhi H, Sadr S,

Babol University of Medical Sciences, Babol, Iran

**Introduction:** Calcium is one of the most important ions for normal function of many organs. But with increase urinary excretion of calcium (hypercalciuria), the risk of renal stone may be increased. Due to difficulty of obtaining a 24h urine (especially in children), a random urine calcium sample is recommended to detect hypercalciuria. However, recent studies have shown that the urinary calcium/creatinine ratio varies with age and geographic areas. So, the purpose of this study was to determine normal values of urinary calcium to creatinine ratio in healthy adolescents' children.

**Materials & Methods:** 480 children aged 12 to 14 years were randomly selected from middle school in Babol (north of Iran) and early morning urine samples of them were studied for determine normal urine Ca / Cr, Na /Cr and K/Cr ratios. Children with family history of renal disease were excluded from this study.

**Results:** In our study, 50% and 95% of urinary Ca/Cr ratio were 0.078 $\pm$ 0.025 and 0.13 mg/mg in the group. The mean of urinary Ca/Cr ratio in boys and girls were 0.079 $\pm$ 0.028 and 0.077 $\pm$ 0/022, respectively. The mean of urinary Na/Cr ratio in boys and girls were 1.39 $\pm$ 0.48 and 1/21 $\pm$ 0/33 respectively. The mean of urinary K/Cr ratio in boys and girls were 0.30 $\pm$ 0.11 and 0.29 $\pm$ 0.10, respectively.

**Conclusions:** This study was shown that urinary Ca/Cr ratio of these children is different from other geographic areas. Also, a direct relationship was seen between urinary Ca/Cr ratio, Na/Cr and k/Cr ratios.

**Tues -O12**

## Hypocitraturia as Second Cause of Infantile Renal Stone in South-West of Iran

Valavi E, Ahmadzadeh A, Amuri P, Alemzadeh-Ansari MH,
Jundishapur University of Medical Sciences, Ahvaz, Iran.

**Introduction:** Renal stones in infants are relatively infrequent but its incidence has increased in recent decades. The aim of this study was to investigate the clinical presentation, metabolic risk factors, and urinary tract abnormalities in infants suffering from renal stone.

**Materials & Methods:** All of 152 infants that were admitted in our department between 2009 and 2012 with ultrasonography proven urolithiasis were included. Unlike previous studies we used foley catheter for accurately collecting of the urine. Ultimately 24-h urine samples were analyzed for calcium, citrate, oxalate, uric acid and magnesium. For detecting cystinuria, qualitative measurement of urinary cystine was done by

nitroprusside test. Also, urinary tract structural abnormalities were evaluated in these patients.
**Results:** The average age at diagnosis of stone was 5.46 months (range, 15 days-12 months). The most common clinical findings in our patients were restlessness and urinary tract infection UTI. Family history of kidney stone found in 67.1% of patients and 68.4% of them were born from consanguineous marriages. Metabolic abnormalities and urinary tract abnormalities were found in 96.1% and 15.1% of children respectively. The most common metabolic risk factors were hypercalciuria (79.6%) and hypocitraturia (40.9%). Hypocitraturia was significantly more common in hypercalciuric and normal uric acid excreted infants (69.4%, *p* value=0.002 and 77.4%, p=0.018 respectively). Hyperoxaluria and hypomagnesuria were found in about 28% of patients and both of them were significantly associated with bilateral urolithiasis (*p*=0.006 and *p*=0.005 respectively). Cystinuria was detected in 3.3% of patients of which 60% were bilateral. Urinary tract abnormalities were diagnosed in 15.1% of patients and was significantly more common in girls (*p*=0.001).
**Conclusions:** Our results showed that urinary metabolic abnormalities especially hypercalciuria and hypocitraturia are very common in infants with urolithiasis; so appropriate evaluation of urinary metabolic parameters can lead us to proper diagnosis and treatment.

**Tues -O13**

## Relation between symptomatic IHC with UTI in patients attending to Children Hospital of Bandarabbas

Ghassemi K, Esteghamati M,

Children Clinical Research Development Center, Hormozgan University of Medical Sciences, Bandarabbas, Iran

**Introduction:** Urinary tract infection (UTI) is known as one of the most frequent diseases in pediatric medicine. A new addition risk factor of predisposing patients to UTI is mentioned as idiopathic hypercalciuria (IHC). IHC is defined as a hypercalciuric status with normal level of calcium in serum and absence of any secondary cause of hypercalciuria. In this study our aim was to evaluate relation between symptomatic IHC with UTI in children.
**Materials & Methods:** This prospective case-control study includes 251 patients who were presented to outpatient clinic of Children hospital of Bandar Abbas. Patients divided in two groups. Case group contains 182 patients with proven UTI and control group contains 69 patients without any signs or symptoms of UTI. Mid-stream urine sample to determine Ca/Cr ratio collected from all patients. Twenty four hours urine collection was ordered for patients with Ca/Cr ration more than 0.21mg/mg. Diagnosis of IHC was based on Ca/Cr ratio more than 4 mg/mg/kg in 24 hours with no secondary cause of IHC.
**Results:** We evaluated association between IHC and UTI in 251 patients. In case group after collecting 24 hours urine, 66 (36.3%) patients and in control group 3 (4.3%) patients detected as IHC. We discovered that frequency of hypercalciuria is higher in female (66.6%) than male (34.4%) and IHC was found significantly more in patients with UTI (P=0.001); but we did not found any evidence to prove relation between recurrent UTI and IHC (P=0.64). Our findings show, the most common manifestations of IHC were dysuria, frequency and hematuria.
**Conclusions:** On the basis of these results, we confirmed the hypothesis that IHC is an important risk factor of UTI (P=0.001) but there was no significant association between recurrent UTI and IHC.

**Tues -O14**

## Evaluation of Predisposing Factors for Nephrolithiasis and Renal Impairment in Patients with Cystic Fibrosis: A preliminary data.

Esfandiar N, Sharifian M, Mohkam M, Dalirani R, Riazi Kh.

Shahid Beheshti University of Medical Sciences, Tehran, Iran

**Introduction:** Patients with cystic fibrosis (CF) are frequently at risks for nephrolithiasis or renal function impairment .For investigation of this complication; renal function tests and metabolic evaluation were performed in these patients.
**Materials & Methods:** Twenty CF patients (10 male, 10 female), mean age 12.3(5-22) years were

enrolled. All patients underwent renal ultrasonography (US) and special blood and 24 hours urinary evaluations.

**Results:** No patient had urolithiasis on US. The mean GFR (Schwartz) was 77.2(range 45.33-143.24 ml/min/1.73m2) and 6/20 patients had GFR<60 ml/min/1.73m2. Hypercalciuria was detected in 8 patients (range1.05-8.05 mg/kg 24h).Hyperuricosuria, hypocitraturia and hyperoxaluria were found in 13(range 3.03-29.25 mg/kg/24h), 11(range 0.23-1.28 mmol/1.73m2/24h) and 2 patients, respectively. Mild proteinuria was observed in 12/20 patients and no one showed proteinuria on urinalysis.

**Conclusions:** There was low level of renal impairment and low molecular weight proteinuria in CF patients. Hypercalciuria, hyperuricosuria, hypocitraturia and hyperoxaluria might lead to the higher risk of urolithiasis in patients with CF.

**Tues -O15**

## Clinical Features and Outcome in Children with Primary Hyperoxaluria: Single Center Experience

Mamizadeh N, Hooman N, Otukesh H, Hoseini R, Nickavar A, Goldasteh M.

Iran University of Medical Sciences, Tehran, Iran

**Introduction:** Primary hyperoxaluria (PHO) consists of enzyme defect of glyoxylate metabolism including AGT and GRHPR .Type 1 is more prevalent than type II or non-type. The aim of this study was to present the clinical presentation and outcome of children with the diagnosis of PHO.

**Materials & Methods:** In this retrospective study, we reviewed data of all patients with diagnosis of PHO who admitted between 2001 and 2012.The criteria of diagnosis were: Recurrent / bilateral/ or multiple urolithiasis; Urine oxalate (mmol/L) to urine creatinin (mmol/L) more than percentile of 95 or 24 hour urine oxalate more than 0.5mmol/1.73/$m^2$; History of calcium oxalate renal stone in relatives or consanguinity of parents; with or without severe chronic kidney disease.

**Results:** From all of patients admitted with the diagnosis of urolithiasis or renal failure, 18patients (12 Females, 6 Males) diagnosed as PHO. The mean age of diagnosis was 4.36 years (range 3 m-13 yrs). The clinical presentations were urolithiasis (n=12) and severe chronic kidney disease (n=6). Five out of six had bilateral nephrocalcinosis and the last one diagnosed by finding oxalate crystals in renal necropsy. The ranges of creatinine at presentation were 0.3 mg/dl to 10.6 mg/dl. From 12 children with urolithiasis, 75% were multiple and 83.3% were bilateral. Failure to thrive was in 44%. Family history was positive for either urolithiasis or hyperoxaluria in 57% and the consanguinity of parents reported in 67%. The patients were followed up for seven years, seven died on CAPD or HD, one is still on HD, four have moderate CKD (creatinin 1.3 to 2.4 mg/dl), three have normal GFR, and three were lost to follow up.

**Conclusions:** The outcome of PHO is poor, high percentage of presentation with renal failure prompt to launch accurate genetic or enzymatic diagnosis in our country to make earlier precise decision for appropriate intensive treatment.

**Tues -O16**

## Neurologic Disorders in Children with Cystinosis

Afsharkhas L

Iran University of Medical Science, Tehran, Iran

**Introduction:** Cystinosis is a metabolic disease characterized by an accumulation of cystine in different organs and tissues, leading to potentially severe organ dysfunction. Three forms of cystinosis have been described: the infantile (nephropathic) form, the intermediate (adolescent, late-onset) form; and the adult (benign) form. Cystinosis is transmitted as an autosomal recessive trait. The gene for nephropathic cystinosis has been mapped to chromosome 17p13 and identified. Now, patients with cystinosis have survived through their fifth decade, but the unremitting accumulation of cystine has created significant non-renal morbidity and mortality. In this present study we investigate neurologic disorders in children with cystinosis.

**Materials & Methods:** In this descriptive, cross sectional study, we evaluated 40 patients with cystinosis referred to neurology clinic in Ali-asghar children hospital between 2008 and 2013. Data was recorded on age, sex, and neurologic disorders.

**Results:** Forty patients with 3-23 years of age had cystinosis. Mean age was 8.65±4.74 years. Twenty cases (50%) were male. Patients had neurologic disorders including seizures (25%), tension type headache (15%), distal muscle weakness (5%), increased ICP (Intracranial pressure) (2.5%), and optic atrophy (2.5%). None of them had swallowing problem. One case died with refractory seizures and increased ICP.
**Conclusions:** Severe neurologic complications such as cerebral atrophy, encephalopathy, increased ICP and progressive myopathy in patients with cystinosis are rare especially in childhood period, after early treatment with Cysteamin, and renal transplantation. Other neurologic disorders should be evaluated and treated properly.

Tues -O17

## The Effect of Phototherapy on Urinary Calcium Excretion Term Neonates with Hyperbilirubinaemia

Safaei-Asl A, Zarkesh M, Hajikhani K,

Guilan University of Medical Sciences, Rasht, Iran

**Introduction:** Phototherapy plays a significant role in the treatment and prevention of hyperbilirubinaemia in neonates. However, this treatment modality may itself result in the development of some complications. A less-known complication of the phototherapy is hypocalcemia. Some studies reported hypocalcemia accompanied by increase in urinary excretion of calcium. The aim of this study was to evaluate the effect of phototherapy on urinary excretion of calcium in term neonates.
**Materials & Methods:** In a before-after study 80 term icteric newborns undergoing phototherapy were selected through accidental sampling. The survey was approved by the ethics committee of Guilan University and informed consent was given by all parents. Neonates who needed antibiotic therapy or blood exchange were excluded. Indication for phototherapy was a serum bilirubin more than 15mg/dl concentration. Continuous phototherapy was used for treatment. Serum samples for calcium, bilirubin and sodium and urine samples for calcium, sodium and creatinine before and after phototherapy (48 hour) were checked. UCa/UCr ratio (mg/mg) was determined. Hypercalciuria was defined by a ratio >0.85. Paired t-test and Wilcoxon test were used for statistically analysis.
**Results:** In this study 80 term newborns (46 male and 34 female) with mean age of 7.01±4.13 days ( 3-26 days), gestational age 38.4±0.54 weeks (38-40 weeks), birth weight 3198±373.2 gr 2500-4420 gr, serum bilirubin 16.54±0.92 mg/dl (15-19 mg/dl) were included. The mean level of serum calcium before and after phototherapy were 9.37mg/ml, 9.25 mg/dl respectively ($p>0.05$). The difference between pre- and post-phototherapy urine calcium levels were found to be statistically significant ($p<0.05$). The mean fractional excretion of sodium and mean of QTC before and after phototherapy had no statistical difference ($p>0.05$).
**Conclusions:** The result of this study showed that phototherapy may increase urinary calcium excretion in term neonates. Further investigation for clarifying the importance of this phenomenon is recommended. A more complex, controlled study has been designed to solve these questions. In conclusion, phototherapy might increase urinary calcium excretion in infants so a further investigation should be conducted on more newborns.

Tues -O18

## The Therapy of Nephropathic Cystinosis

Niaudet P. Ptof.

Hôpital Necker - Enfants Malades, Paris, France

**Supportive treatment:** The water intake must be adjusted to diuresis and weight variations. Fluid requirement increases with external temperature and with fever. It is also increased by the required mineral supplements.
Sodium and potassium bicarbonate must be given in order to obtain a plasma bicarbonate level between 21 and 24 mmol/l. This is sometimes difficult and may require large amounts of buffer, up to 10–15 mmol/kg. Hypokalemia requires 4 to 10 mmol/kg potassium supplements in order to maintain serum potassium above 3 mmol/l. Hypophosphatemia must be corrected with a supplement of sodium/potassium phosphate at a dose of 0.3–1 g/day. Excessive phosphorus prescription may lead to nephrocalcinosis. Since tubular 1α□-hydroxylation is diminished in this

disease, it is justified to give 1α- or 1α-25-$OHD_3$ (0.10–0.50 μg/day), especially in cases of symptomatic rickets. Carnitine is given at a dose of 100 mg/kg per day. Feeding problems may require tube or gastric button feeding. Urinary losses may be reduced by the prescription of indomethacin at a dose of 1.5–3 mg/kg. It has been shown that the angiotensin converting enzyme (ACE) inhibitor, enalapril, diminishes albuminuria and possibly slows down the degradation of renal function.

Hypothyroidism, even if asymptomatic, should be treated with L-thyroxine supplementation. Growth failure is improved by administration of recombinant growth hormone at a dose of 1 U/kg/week.

**Specific Therapy:** Cysteamine (Cystagon) prevents cystine accumulation in various organs. The dose is progressively increased from 10 to 50 mg/kg per day. The effect, assessed by cystine assay in leukocytes lasts no longer than 6 h. Consequently, it has to be given in 4 separate doses – one every 6 hours . It was recently shown that a twice daily administration of an enteric release formulation of cysteamine bitartrate was as effective as the current formulation of cysteamine. The aim is to keep cystine content under 2, or better, under 1 nmol of 1/2 cystine per mg of protein. The drug should be started as soon as the diagnosis is confirmed. Side effects of the drug include nausea and vomiting and can be managed with omeprazole. Less commonly, allergic rashes, seizures and neutropenia are seen. In addition, cysteamine is responsible for an unpleasant breath odour so that compliance with 4 doses per day is difficult to maintain in the long term, especially in adolescents. Cysteamine eye drops are able to prevent corneal deposits, and may decrease and even suppress the deposits already present.A new therapeutic approach was tested on the animal model using bone marrow cell transplantation with encouraging results

## First day Poster Presentations

### Tues - P1

### Fanconi Syndrome: Review of Diagnosis and Management in Patients- Single Center Report

Ghasemian A, Hooman N, Otukesh H, Hoseini R, Nickavar A,

Iran University of Medical Sciences, Tehran, Iran

**Introduction:** Fanconi syndrome is a generalized inherited or acquired disturbance of renal tubular transport. The aim of the present study was retrospectively review of the initial clinical feature, laboratory tests disturbance, and management.

**Materials & Methods:** We retrospectively reviewed 14 patients who admitted in Ali-Asghar children hospital between March 2003 and May 2013. Inclusion criteria were the presence of proximal renal tubular acidosis (normal anion gap hyperchloremic metabolic acidosis, urine pH less than 5.6), glycosuria (positive urine dipstick for glucose), generalized aminoaciduria, normal blood aminoacid chromaography, phosphaturia (fractional excretion of Phosphate more than 25%), and rickets.

**Results:** From all children with diagnosis of renal tubular acidosis, 14 had Fanconi syndrome, 20 Von –Gierke, Fanconi Bickel in four, galactosemia in 22, and tyrosinemia in 9. Of 14 patient, 8 (57%) were females and 6 (43%) males. The mean age of children was 9.1 years (range: 3 -18 years). The mean age of patients at presentation was 7 months (range: 3-11 months). Consanguinity reported in 12 (85.7%) children. The most frequent manifestations were impaired growth (92.8%), polyuria and polydypsia (78.6%), dehydration (64.3%), and rickets (57.2%). All of the patients had a normal anion gap renal tubular acidosis, aminoaciduria, glycosuria, bicarbonaturia, phosphaturia, hypophosphatemia, and hypokalemia.

All children received citrate solution, fluid, phosphate, and vitamin D.

Conclusion: The low rate of idiopathic Fanconi syndrome urges the evaluation for secondary causes of Fanconi syndrome.

### Tues - P2

### Pseudohypoaldosteronism in Iranian Children-Case Series

Ahmadi B, Razzaghy-Azar M, Khalesi N,

Iran University of Medical Sciences, Tehran, Iran

**Introduction:** Pseudohypoaldosteronism is a rare disease that characterized by low Na serum and

elevated K serum. We are going to report clinical and laboratory finding of six patients.

**Materials & Methods:** This is a case series study. The data of children, who were admitted between 2005-2009 in Ali-Asghar children hospital, have been reviewed. Inclusion criteria were hypoaldosteronism and hyperreninemia in the presence of hyponatremia and hyperkalemia.

Demography data, laboratory tests, treatment, and outcome have been recorded.

**Results:** Six patients (5 males and 1 female) with median age of 2 months (range 10 day-5.5 years) had sufficient criteria. Median body weight at presentation was 3.6 Kg (range 2.95-4.55).. All patients admitted with poor feeding and failure to gain weight. Familial history was positive in only two patients. All of the patients went under treatment with hydrocortisone and standard medical treatment for hyperkalemia. One patient died and other discharged with maintenance medical therapy. Outcome: All patients have been followed for median of 5.5 years and except one, the rest of 5 patients are still coming for follow up in our.

**Tues - P3**

## Barrter Syndrome and Sensorineural Deafness: A Case Report of Type IV Barrter Syndrome

Mortazavi F,

Tabriz University of Medical Sciences Tabriz, Iran

**Introduction:** Barrter syndrome type IV is a rare autosomal recessive disease characterized by hypokalemic metabolic alkalosis, severe renal salt wasting and sensorineural hearing loss without nephrocalcinosis. Its pathophysiology is dysfunction of barttin which is an accessory subunit of chloride channels located in thick ascending loop of henle, descending convoluted tubules of kidney and inner ear. Barttin modulates the stability, cell surface localization and function of ClC-K channels. Various mutations of causal gen, BSN, lead to phenotypes of varying severity. We report a case of this disease from Children's Hospital of Tabriz.

**Case report:** A 50- days old girl with 2 kg weight was admitted in surgery ward with vomiting and abdominal distention with probability of Hirschsprung's disease. She had a history of premature birth at 34 weeks of gestational age. Laboratory tests revealed severe hyponatremia (113 meq/L) and hypokalemic metabolic alkalosis. Serum chloride was 91 and urine chloride was 115 meq/L. Imaging studies for Hirschsprung's disease, renal ultrasonography, and sweet test were normal. In nephrology consult diagnosis of Barrter syndrome was made and she was discharged with K-Cl tablets and Indomethacin capsule. At 1.5 years of old, parents noted hearing difficulty and she underwent insertion of bilateral Ventilation Tube and adenoidectomy with diagnosis of refractory serous otitis media. However, hearing impairment was continued after surgery. Auditory brain stem response (ABR) and Otoacoustic emission (OAE) tests showed a sensorineural deafness and now the patient is a candidate for Cochlear implantation.

**Conclusion:** All patients of Bartter syndrome should be evaluated for sensorineural deafness in infancy because undiagnosed deafness may have detrimental effect on development of the child.

**Tues - P4**

## Senior-Loken Syndrome, Role of Cilia in Tubulopathies.

Momtaz HE,

Hamadan University of Medical Sciences, Hamadan, IRAN

**Introduction:** Nephronophthisis is an autosomal recessive cystic renal disease and is supposed to be the most common genetic cause of ESRD in first 3 decades of life. This disease can be present in infancy, adolescence (juvenile type) and adulthood. In rare instances it may be associated with extrarenal involvement such as retinitis pigmentosa (Senior-Loken syndrome) and cerebellum (Joubert syndrome). Recently, role of ciliary dysfunction is emphasized in many diseases and syndromes which their manifestations previously seemed to be not related to each other at all. In "Ciliopathies" kidney involvement presents as cystic or dysplastic renal disease resulting in ESRD eventually, eye involvement in form of retinitis pigmentosa can cause blindness, cerebellar involvement causes nystagmus and ataxia and so on. Here we present a case of Senior-Loken

syndrome with ESRD and her brother with only ophthalmic involvement.

**Case Report:** A 13- year old female with past history of blindness presented with pallor, pain on lower extremities and polydypsia. Blood pressure was normal. Initial laboratory investigation showed: Hb= 7.5 g/dl, BUN=33 mg/dl, and creatinine = 2.8 mg/dl. Urinalysis was normal. Parents were relatives (cousins) and she had younger 6- year old brother with severe visual impairment but his renal function was within normal limit. Ophthalmologic examination of our case was in favor of retinitis pigmentosa. Renal biopsy findings was suggestive of juvenile nephronophthisis with tubular atrophy, thickened tubular basement membrane , interstitial fibrosis and glomerulosclerosis. Because of these clinical and pathological findings she was labeled as a case of Senior -Loken syndrome. During follow- up, her creatinine rose up and she underwent hemodialysis after her family rejected peritoneal dialysis as a renal replacement option and now she is preparing for renal transplantation. Literature search about this case gave valuable information about "Ciliopathies", a group of previously supposed unrelated organ involvement in various syndromes which now could be described as ciliary dysfunction in kidney, eye, ear, adipose tissue, CNS and probably other organs. In kidneys ciliopathy results in abnormal tubular development, cystic and dysplastic changes that may ultimately deteriorate to end stage renal disease.

**Conclusions:** Multiple organ involvement in a patient with chronic kidney disease should raise suspicion of possible "ciliopathy" syndromes and because of strong probability of autosomal recessive and dominant inheritance, all first degree relatives should be screened and followed for renal involvement.

**Tues - P5**

## Carnitine Deficiency in Chronic Kidney Disease Stage V: Comparing Hemodialysis with Peritoneal Dialysis Subjects

Naseri M, Esmaeeli M, Ghaneh- Sherbaf F, Vakili V, Rasoli Z, Jahanshi SH,

Mashhad University of Medical Sciences, Mashhad, Iran

**Introduction:** Carnitine deficiency is common in chronic hemodialysis patients. This study aims to define the frequency of Carnitine deficiency with considering the effects of age, gender, duration and modality of dialysis.

**Materials & Methods:** Forty seven dialysis patients including 20 girls (42.5%) and 27 boys (57.5%), aged 19-300 (166.02 ±76.09) months including 13 (31.7%) CAPD and 28(68.3%) hemodialysis cases enrolled the study. Times from onset of dialysis were 1to128 months (mean: 44.28±31.26SD). The deficiency was defined as serum levels of free carnitine less than 7 µmol/l and plasma levels of Acylcarnitine below 15 µmol/l. Patients divided into two groups: Patients with normal and those with low plasma levels. Chi square and student T tests were used. P values <0.05 were considered as statistically significant differences.

**Results:** Serum levels of free carnitine were normal in 45(95.7%) and high in 2(4.3%) patients. Total serum carnitine levels were normal in 44(93.6%) and high in 3(6.4%) subjects. Plasma levels of Acylcarnitine were low in 23(48.9%) and normal in 24(51.1%) of enrolled cases. There was no significant difference in frequencies of Acylcarnitine deficiency in hemodialysis versus peritoneal dialysis subjects (P=0.135), girls versus boys (P=0.76), mean ages of patients (P=0.179), and dialysis duration (P=0.126) between groups. Twenty two patients received oral carnitine 250-1000mg/day, while 13 did not receive the drugs. Low plasma levels of Acylcarnitine were reported in 11 and 6 subjects, respectively (P>0.05).

**Conclusions:** Acylcarnitine deficiency was common in our series. The deficiency was as common in hemodialysis as peritoneal dialysis cases. Although in normal population the deficiency is greater in females; in dialysis patients the deficiency was as common in boys as girls.

**Tues - P6**

## Association Study of TLR4 (Asp299Gly, Thr399Ile) Gene Polymorphisms in the Severity (Amyloidosis) of Familial Mediterranean fever in North-West of Iran

Laghaeian N.[1], Bonyadi M.[2], Abedi Azar S.[3]

[1]Ahar Branch of Islamic Azad University, [2] Center of Excellence for Biodiversity, Faculty of Natural Sciences, University of Tabriz,.[3] Tabriz University of Medical Sciences, Tabriz, Iran

**Introduction:** Familial Mediterranean Fever (FMF) is an autosomal recessive auto-inflammatory disorder with more than 60 disease-associated mutations in the responsible gene, *MEFV*. Amyloidosis is the major complication of Familial Mediterranean Fever. A less frequent but most severe complication of FMF is the development of renal amyloidosis, ultimately leading to end-stage kidney failure. Toll-like receptors (TLR) are involved in the activation of an innate immune system. TLR-4(Asp299Gly and Thre399Ile) polymorphisms down-regulate inflammation. We investigated the effect of these polymorphisms on the development or resistance of amyloidosis in FMF patients.

**Materials & Methods:** In this study we investigated 86 FMF patients (27 with amyloidosis) and 66 matched control subjects. TLR-4(Asp299Gly and Thr399Ile) polymorphisms were analyzed with the polymerase chain reaction restriction fragment length polymorphism method (PCR-RFLP).

**Results:** The frequency of these polymorphisms were not different in FMF patients (with or without amyloidosis) compared to the control group. Comparison between FMF patients and control subjects revealed no significant association of TLR-4(Asp299Gly and Thre399Ile) polymorphisms, however significant associations between two groups of patients with specific symptoms were observed.

**Conclusions:** Previous studies suggest the association between TLR-4(Asp299Gly and Thr399Ile) polymorphisms and severity of FMF; However results of present study demonstrated no association between these polymorphisms and frequency of attacks or development of amyloidosis.

**Tues – P7**

## Distal Renal Tubular Acidosis Associated with Hemihypertrophy (Case Report)

Ghassemi K, Esteghamati M

Children Clinical Research Development Center, Hormozgan University of Medical Sciences, Bandarabbas, Iran

**Introduction:** Primary distal renal tubular acidosis(dRTA) is an inherited disease characterized by a normal serum anion gap (AG) with hyperchloremic metabolic acidosis (HClMAC.), high urinary pH(UpH), growth retardation, nephrocalcinosis (NC), and renal stone.

**Case report:** A 6-months boy was referred to pediatric nephrology clinic with history of hemihypertrophy of right side of body and failure to thrive(FTT) and bilateral nephrolithiasis (NL) without NC He was a full term baby with a birth weight of 2.85 Kg and height of 49 cm. His parents were not first degree relatives and family history was negative for renal stone. His physical examination revealed a weight of 4.5 kg and height of 54 cm at age 6 mon. Initial laboratory investigations showed HClMAC. [Cl=119 mmol/L, PH=7.26, $HCO_3$= 13.7, BE = 11.4mmol/L] with hypokalemia (hypoK), normal serum AG):11meq/L, and positive urine AG: 4 meq/L. Random Uca/Ucr was 1 and 24- hour Uca 5 mg/kg/day. Other hormonal profile such as FT4, TSH, LH, FSH, prolactin, and growth hormone level were normal. Serum phosphate: 5.6 mg/dl, Ca: 8.6 mg/dl, ALKP (350 IU/L), total serum protein:60 mg/dl, Cr:0.4 mg/dl, and UpH: 6.3 . The ultrasound scan of patient revealed NL without NC and normal position of both kidneys.

**Conclusions:** Results presented with unusual clinical feature (hemihypertrophy) of this disease described so far. Distal RTA is a rare renal disorder characterized by normal AG - HClMAC. and hypoK, UpH consistently above 5.5, hypercalciuria, hypocitraturia, potassium wasting in urine, NL, and NC. Our patient is an atypical presentation of dRTA. Renal acidification defect with hypercalciuria and hypocitrturia are important factors in the pathogenesis of nephrolithiasis Urinary citrate are an inhibitor of crystal aggregation and precipitation. When citrate excretion is reduced, more calcium to be chelated, thus renal stone develops. RTA may be accompanied by clinically hypoK due to renal potassium wasting which leads to impaired growth and FTT. It is important to differentiate primary causes of FTT from secondary causes such as d RTA and also hemihyperthrophy associated with neurofibromatosis and vascular

disease may be an associated finding with other disorders such as d RTA as a rare and new report.

**Tues – P8**

## Novel Mutations in PKD1 Gene in An Iranian Patients Family With Autosomal-Dominant Polycystic Kidney Disease

Hafizi A[1], Khatami SR[1], Galehdari H[1], Shariati GR[2], Saberi AH[3], Hamid M[5]

[1] Shahid Chamran university of Ahvaz, Iran;[2] Narges genetic Laboratory of Ahvaz, [3]Jundishapur University of Medical Science, Ahvaz, Iran, [5]Research center of Biotechnology Anstitute Pastor, Tehran, Iran

**Introduction:** Autosomal dominant polycystic kidney disease (ADPKD) is one of the most common genetic kidney disorders with the incidence of 1 in 1000 births. ADPKD is genetically heterogeneous, with two genes identified: PKD1 (16p13.3, 46 exons) and PKD2 (4q21, 15 exons). 85% of patients with ADPKD have at least one mutation in the PKD1 gene. Genetic studies have demonstrated an important allelic variability among patients but very few data are known about the genetic variation in Iranian populations.
**Materials & Methods:** In this study, coding exons analysis of PKD1 by exon direct sequencing was performed in a 7-year old boy with ADPKD and his parent. The patient's father is ADPKD affected without any kidney dysfunction and the patient's mother is congenitally missing one kidney
Results: Molecular genetic testing fund a doubted pathogenic mutation in all 3 members of this family. It was a missense mutation GTG>ATG at position 3057 in exon 25 of PKD1. On the other hand two Novel missense mutations were reported, ACA>GCA found in exon 11 at codon 2241 and CAC>AAC found in exon 38 at codon 3710.
**Conclusions:** GTG>ATG causes the conversion of amino acids V to M. then for checking the validation of the software reports based on the pathogenicity of this mutation, exon 25 of 50 unrelated normal cases matched by sex and Ethnicity were sequenced. Our findings suggested that this mutation is a polymorphism with high frequency (60%) in the population of south west Iran. The second and third mutations were Novel polymorphisms that changed Threonine to Alanine at codon 2241 and Histidine to Asparagine at codon 3710.