Abstracts

Background FHHNC is a rare autosomal recessive tubulopathy of the thick ascending limb due to inactivating mutations in the Claudin-té and Claudin-tó genes which are responsible for the paracellular reabsorption of calcium and magnesium. Clinically, FHHNC is characterized by urinary tract infections, nephrolithiasis, nephrocalcinosis and progressive renal failure.

Objective To present clinical and molecular data on 2 siblings with FHHNC and early onset SND, found to have a new mutation in the Claudin-té gene.

Methods A 16-year-old male and his 14-year-old sister were diagnosed with chronic kidney disease since early infancy, manifested by recurrent UTIs, polyuria and polydipsia, poor growth, mild mental retardation and delayed speech. SND was diagnosed at the age of 6 and 7 years. Both have hypomagnesemia, hypermagnesuria, hypercalcemia and nephrocalcinosis. Genotyping was performed by PCR using tetranucleotide repeat polymorphisms. Specific primer pairs of genomic DNA were used as template for sequencing the Cldn16 gene.

Results Exons 1, 2, 4 and 5 were amplified and revealed wild type sequencing but exon 3 failed in amplification by PCR. Long range PCR spanning exon 2 to 4 of the gene yielded a 2.5 kb fragment shared by the patients. Sequencing of this fragment reveal a 2630 bp shift of Cldn16 protein.

Conclusion This family demonstrates the first identified homozygous mutation in the Claudin-té gene causing the deletion of an entire exon. This, however, is unlikely to explain the SND, since this gene is not expressed in the cochlea.

PROTEOLYTICAL ENZYMES IN GLOMERULONEPHRITIS

doi:10.1136/archdischild-2012-302724.1202


Aim of the investigation: Study of activity of proteolytic enzymes in children with acute (AGN) and chronic (CGN) glomerulonephritis.

Materials and Methods Were investigated 50 children with acute and chronic glomerulonephritis, age from 7 to 15 years, including 30 children with AGN plus nephrotic syndrome, and 20 children with CGN, nephrotic form, period of activation and remission. Control group including 20 practically healthy children. In serum determined activity of trypsin, a-antitrypsin, and a2-macroglobuline.

Results In AGN with nephrotic syndrome, at the initial stage of the process, as well as in activation of CGN, were determined significant increasing of trypsin, a-antitrypsin, and a2-macroglobuline in comparison to control group (P<0.01). The activity of a2-macroglobuline in children with AGN plus nephrotic syndrome and CGN, nephrotic form, period of activation and remission. Control group included 20 practically healthy children. In serum determined activity of trypsin, a-antitrypsin, and a2-macroglobuline in comparison to control group (P<0.01). The activity of a2-macroglobuline in children with AGN plus nephrotic syndrome was 11.9±1.42 g/l, P<0.001, what 1.9 fold higher than in control group (6.2±0.030 g/l). In activation of CGN, nephrotic form, the activity of a2-macroglobuline was in 1.5 fold higher the indexes of control group (9.5±0.90 g/l, P<0.01).

In remission, the values of trypsin, a-antitrypsin, and a2-macroglobuline decreasing, but did not attain the basic level of control group, which suggests the persistence of activity of pathological process in kidneys.

Conclusion Determining of activity of proteolytic enzymes in serum in diagnostic criterion for determining of severity, activity of pathological process in kidneys, and determining the outcome the complications of glomerulonephritis.

THE DIABETIC PREGNANCY AND OFFSPRING BLOOD PRESSURE IN CHILDHOOD: A SYSTEMATIC REVIEW AND META-ANALYSIS

doi:10.1136/archdischild-2012-302724.1203

Background and Aims Offspring of diabetic mothers (ODM) are at increased risk of the metabolic syndrome in later life. We aimed to perform a systematic review and meta-analysis of studies examining offspring systolic and diastolic blood pressure (SBP, DBP) in childhood in relation to maternal diabetes.

Methods Citations were identified in PubMed. Authors were contacted for additional data where necessary. SBP and DBP in ODM and controls were compared. Subgroup analysis was performed according to type of maternal diabetes and offspring gender. A fixed effect meta-analysis was performed, and a random effects analysis where significant heterogeneity was present. Meta-regression was used to test the relationship between offspring SBP and maternal pre-pregnancy BMI.

Results Fifteen studies were included in the systematic review and thirteen in the meta-analysis. SBP was 1.88mmHg higher in ODM (95% CI 0.47, 3.28; p=0.009). The increase in SBP was similar in both offspring of mothers with gestational diabetes (1.39mmHg [0.00, 2.77]; p=0.05) and type 1 diabetes (1.64mmHg [0.09, 3.18]; p=0.04). Male ODM had higher SBP (2.01mmHg [0.93, 3.10]; p=0.0003) and DBP (1.12mmHg [0.36, 1.88]; p=0.004) than controls, but the differences in SBP and DBP between female ODM and controls were not statistically significant. Offspring SBP was positively correlated with maternal pre-pregnancy BMI; however, the association was not significant (p=0.37).

Utility of Serum Procalcitonin to Predict Renal Scars in Infant with Fibrile Urinary Tract Infection

doi:10.1136/archdischild-2012-302724.1204

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Background and Aims The aim of this study was to evaluate the usefulness of Procalcitonin (PCT) as a marker for renal scars in infants with a first febrile urinary tract infection (UTI).

DMSA scintigraphy, the gold standard for detection of cortical scarring, has clearly shown that not all febrile UTI are associated with renal lesions and that common clinical and laboratory evaluations are not reliable to distinguish between acute pyelonephritis (APN) and simple UTI. Scarring secondary to APN is a common event occurring in approximately 30% of all cases.

PCT, prohormone of calcitonin has been measured in various systemic inflammatory response syndromes, because it appears to be correlated with the severity of microbial invasion and it can be used to check for the presence of parenquimal scars.

Conclusions ODM have higher SBP than controls. This increase is independent of type of maternal diabetes and may be related to maternal pre-pregnancy BMI. Gender-specific differences require further investigation.