Pediatric Nephrology 2015 **30:9** (1679)

Recurrent pancreatitis in a girl with WT1 mutation

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Abstract

Introduction: WT1 mutations cause a wide spectrum of renal and extrarenal manifestations. Pancreatitis in childhood is a plurifactorial disease which can be caused by toxic, metabolic, infectious, genetic or anatomic factors. Case description: A 1-month-old girl presented with edema ,extreme hyponatriema and proteinuria. Due to rapid progression to terminale renal failure , renal replacement therapy was necessary and initially peritoneal dialysis was started. Kidney biopsy revealed diffuse mesangial sclerosis and genetic analysis revealed a de novo heterozygous mutation c.1323C>G or p. His441Gln substitution within the zinc finger-2 domain of the WT1 gene, confirming the diagnosis. Unless application of frequent exchanges with Physioneal PD solution(Baxter) there was insufficient clearance of the large molecules and ultrafiltration. Icodextrine (Extraneal Baxter) during the day was associated. she supported this regimen well and gained weight. However at the age of 16 months she was admitted with a first episode of severe acute abdominal pain. There were no signs of peritonitis. Biochemical investigations could not reveal any abnormality and ultrasound of the abdomen was normal. Explorative laparotomy was negative. No diagnosis could be made at that time. Unfortunately one month later she developed a new episode of extreme acute pain crisis. Lipase was extreme high (1384 U/L) and ultrasound revealed signs of pancreatitis. Additional investigations could not detect any infection or anatomic abnormality with obstructive pathology. She used no toxic medications. In literature there were some case reports (in adults) about the association between the use of icodextrine and pancreatitis. The use of this peritoneal solution was stopped. Nevertheless she continued to experience recurrent episodes of severe painfull pancreatitis with a frequency of 1/8 weeks. Except for one episode (after vaccination) no other triggers were identified. We decided to stop treatment with peritoneal dialysis and started with hemodialysis. Since then one pancreatitis episode / 6 months occurred. Genetic analysis of SPINK1,PRSS1 and CFTR-gen did not show any mutation. Conclusions: This case shows an unusual association between a congenital

nephropathy and recurrent pancreatitis. We hypothesize that this girl developed a first viral pancreatitis which lead to a severe inflammation of pancreas which made her vulnerable to recurrent episodes in addition to peritoneal dialysis, hypercalcemia and other viral triggers(HZV, CMV). Or the phenotypic expression of the recurrent pancreatitis in this girl might be due to an additional genetic modifier , associated with the known genetic mutation in WT1 gen.

Drug terms

peritoneal dialysis fluid zinc finger protein triacylglycerol lipase

Disease terms

pancreatitis kidney failure kidney disease inflammation hypercalcemia proteinuria edema infection pain sclerosis peritonitis acute abdomen

Other terms

girl **female** **human** **mutation** **European** **society** **nephrology** peritoneal dialysis ultrasound genetic analysis diagnosis renal replacement therapy pancreas hemodialysis vaccination ultrafiltration adult gene laparotomy abdomen kidney biopsy pathology drug therapy case report weight childhood

Additional information

Publication type	Conference Abstract
Embase identification number (PUI)	L72033583
Digital Object Identifier (DOI)	10.1007/s00467-015-3158-7
Entry date	2015-10-17 (Full record)
Source of indexing	Embase
Page range	1679 -
Language of article	English

Language of summary	English
Source type	Journal
Abbreviated journal title	Pediatr. Nephrol.
Source publication date	September 2015
ISSN	0931041X (print)
Conference name	48th Annual Scientific Meeting of the European Society for Paediatric Nephrology, ESPN 2015
Conference date	2015-09-03 to 2015-09-05
Conference location	Brussels, Belgium
Country of author	Belgium

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