right side), without organic obstruction. This is a serious condition that can be complicated by perforation, peritonitis and death. The underlying cause is multifactorial (altered regulation of the intestinal sympathetic – parasympathetic system, among other factors), and treatment is fundamented on endoscopic decompression.[4] It has been reported that certain drugs such as the calcium antagonists used by our patient (amlodipine) aggravate intestinal dilatation.

In our case we observed all the described complications following the intestinal infusion of P-rich enemas – including hypernatremia, possibly due to increased Na absorption, fluid sequestration in the bowel lumen, and volume depletion causing hypertonic dehydration with functional renal failure. Hyperphosphoremia is associated with hypocalcemia, tetanic contractions and extraskeletal calcium phosphate deposits. Hemodialysis is the treatment of choice.[5] In our patient, a single session sufficed to improve the condition, though the medical treatment provided must have contributed to such improvement. Lastly, it should be pointed out that there are other types of enemas that do not contain P and which can be administered to patients with renal failure, such as X-Prep® or Puntualex® (sennosides A and B).

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Compound heterozygosis for intrón 9 + 1 g > T and Leu 850pro mutations in the SLC12A3 gene in Gitelman's syndrome

Nefrología 2008; 28 **(6)** 659-660

To the editor: Gitelman's syndrome is the most frequent hereditary tubular disease in Europe (1% of heterozygous individuals),[1] and is characterized by hypopotassemia, metabolic alkalosis, hypomagnesemia and hypocalciuria.[2] A recessive autosomal hereditary pattern is observed, related to mutations of the SLC12A3 gene, which encodes for the Na/Cl co-transporter in the renal distal tubule, causing sodium reabsorption failure and secondary hyperaldosteronism. Over 100 different mutations have been reported.

CLINICAL CASE

A 35-year-old male presented with a history of generalized weakness in the course of an episode of acute diarrhea. Plasma potassium concentration was 1.1 mEq/l. Upon readmission to a tertiary hospital due to the relapse of severe and symptomatic hypopotassemia, the laboratory tests revealed the presence of full Gitelman's syndrome (table I). A study of the family detected a similar biochemical condition only in the male sibling of the patient; both individuals had suffered scant previous repercussions in the form of carpopedal spasms in childhood, in relation to fever episodes, and occasionally also in adult life. Treatment was provided in the form of potassium and magnesium supplements, spironolactone, amiloride and finally eplerenone, at very high doses for normalization of the plasma levels – with the persistence of important potassium elimination in urine. The genetic study showed both siblings to carry the int. 9 + 1G > T mutation, present in the father, and the Leu850Pro mutation in exon 22, present in the mother. The two children of the patient carried only the Leu850Pro mutation.

DISCUSSION

The association of these two mutations is reported for the first time. The variant int. 9 + 1G > T has been found in the largest published series with a common mutation: 12 gypsy families with no relation to each other.[3] In this group, the affected individuals were homozygous, while the parents and descendents were healthy heterozygous carriers. This points to the possible reinforcing effect of the second mutation (Leu850Pro) upon the first, since ours are the first patients described with clinical involvement in which the variant int. 9 + 1G > T is present in heterozygosis. All patients with clinical manifestations in which a single mutation is detected should be regarded as compound heterozygotes in which the second mutation has not been detected, due to incomplete sequencing of the gene. The presence of compound heterozygosis suggests that the transporter alteration requires at least two variations in order to

Table I. Representative blood and urine test parameters of the original patient, upon tertiary hospital admission. The normal values, corresponding to body weight, appear in parentheses. The transtubular potassium gradient was 36

Plasma Mg	1.11 mg/dl	(1.4-2.7)
Plasma K	2.3 mEq/l	(3.5-5.1)
Urine K	81 mEq/l	
Urine Ca	116 mg/24 h	(350-360 mg/24 h)
Plasma renin activity	29.7	(0.2-2.3)
Plasma aldosterone	1,256.64 pg/ml	(40-300)

manifest, and which could determine a variety of structural changes with different effects upon transporter activity – thus giving rise to clinical conditions of different degrees of severity.[4]

It should be pointed out that the diagnosis is established in adult life, with scant prior clinical expression. This contrast with the severity of hypopotassemia and hypomagnesemia, and the usually intense physical activity of the patients – reflecting the scant correlation found in most cases between the biochemical alterations and the clinical picture.[5] Therefore, this diagnosis must be considered in all cases of hypopotassemia with inappropriately high potassium elimination in urine, in the absence of arterial hypertension. The condition was initially not suspected, despite the recording of a plasma potassium concentration of 1 mEq/l (infrequent even in severe diarrhea), because of the lack of a urinary potassium measurement that constitutes a key element in the diagnosis.

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Extravascular misplacement of the tunneled hemodialysis catheter

Nefrología 2008; 28 **(6)** 660-661

To the editor: The use of central venous catheters for long-term vascular access is an increasingly common practice. The most frequent complications associated with the use of such catheters include aspects relating to their insertion.[1]

We report two complications of tunneled catheter placement. In both cases the catheter was successfully removed in the operating room.

CASE 1

A 58-year-old male presented with chronic kidney failure secondary to diabetic nephropathy, subjected to hemodialysis since April 2006 through a tunneled catheter inserted in the right innominate vein. The patient was admitted to our Service on December 20, 2006, due to sepsis probably related with the catheter. Despite antibiotic treatment, the fever failed to subside, and the catheter was therefore removed. Following clinical improvement of the patient, an attempt was made to tunnel a new catheter to the right innominate vein. The vein was punctured, and after drawing blood of venous appearance, the catheter was positioned. The catheter failed to function, however, and chest X-rays revealed an extravascular position of the tip, with a right pneumothorax. Withdrawal in the operating room was decided jointly with the Services of Chest Surgery and Vascular Surgery.

Following contrast administration via the axillary vein, the catheter was seen to traverse the subclavian vein (fig. 1a). Removal of the former was then carried out under imaging control. The post-withdrawal radiological control revealed linear contrast leakage tracing the cathe-

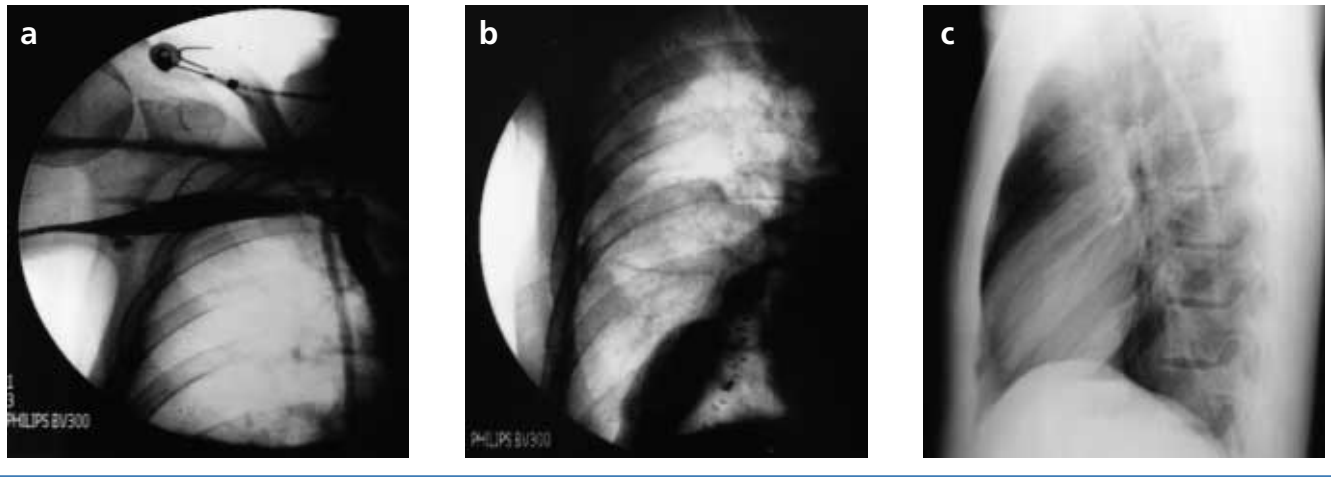

Figure 1. a) Catheter traversing the subclavian vein; b) Contrast leakage following withdrawal; c) Radiological control after catheter insertion in the right innominate vein.