Hypochloremia and hyponatremia as the initial presentation of cystic fibrosis in three adults

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Résumé en anglais	Cystic fibrosis (CF) is caused by mutations in the cystic fibrosis transmembrane conductance regulator (CFTR) gene. Most diagnoses of CF are made during infancy or childhood, and are based on respiratory or digestive involvement. Initial extracellular dehydration leading to the diagnosis of CF is usual in infants but has only exceptionally been reported in adults. We describe three new adult cases of CF initially presenting with depletive hyponatremia and hypochloremia following exposure to heat. At first consultation, these patients had no symptoms suggestive of CF. One patient presented with a seizure induced by hyponatremia. The two other patients were siblings carrying a novel c.4434insA mutation in exon 24 of CFTR. Acute dehydration is a very rare initial manifestation of CF but may be life-threatening. The possibility of CF should not be ignored in cases of depletive hyponatremia, hypochloremia or hypokalemic metabolic alkalosis, even in otherwise healthy patients.
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