Severe episodes of extra cellular dehydration : an atypical adult presentation of cystic fibrosis

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Résumé en anglais	Cystic fibrosis (CF) is usually diagnosed during childhood by respiratory or gastro-intestinal symptoms. Hyponatremic hypochloremic dehydration with metabolic alkalosis is a rare but typical presentation of CF in infants. In contrast, only 3 cases have been described in adults. We report a case of CF in a 33-year-old Caucasian female presenting with a severe sodium and chloride depletion caused by inappropriate sweating. She experienced three episodes of severe dehydration before the diagnosis was suspected. Sweat chloride test was pathological and mild pulmonary involvement was found on CT scan. AF508 mutation and a rare mutation (3849+40 A/G) on the intron 19 of CFTR gene were found. Interestingly, our patient has a heterozygote twin sister, carrier of the same mutations of CFTR gene who also developed CF but with a different phenotype. We suspect modifier genes to be implicated in the differences observed between the two phenotypes. We discuss the physiopathology of electrolyte disturbance and review the other similar adults cases.
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