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Hidden in Plain Sight—A Rare Presentation of Cystic Fibrosis with Pseudo-Bartter Syndrome

Dear Editor,

A three-month-old girl presented with persistent vomiting, failure to thrive and hearing loss. She was static at the birth weight of 3 kg. Investigations revealed hyponatremia (122 meq/L), hypokalemia (2.6 meq/L), hypochloremia (88 meq/L), hypocalcemia (6.8 mg%), and metabolic alkalosis (PH 7.6). Urine electrolytes showed low urine sodium and chloride (10.4 mmol/L and 47 mmol/L, respectively). Brainstem evoked response audiometry (BERA) confirmed bilateral severe hearing loss. Based on the above laboratory findings with sensorineural hearing loss, a diagnosis of Type 4A antenatal Bartter syndrome was made. Indomethacin was planned. A clinical exome test uncovered a homozygous mutation in the CFTR gene on exon 19, with the pathogenic variant of c3107C > T, consistent with the diagnosis of CF. The missense mutation has caused substitution of isoleucine for threonine at codon 1036. This was previously noted in a study done on Iranian patients and was described as a novel mutation.[1] She is being treated with supplements of salt and calcium, and cochlear implant is planned.

The incidence of pseudo bartter syndrome (PBS) in CF ranges from 12% - 16%.[2] Persistent vomiting is a distinctive warning sign for early identification. The mechanism is activation of renin angiotensin aldosterone system due to excessive loss of sodium chloride in sweat. This is in contrast to Bartter syndrome, where there is defective sodium reabsorption from the thick ascending limb of Henle.[3] PBS results in low levels of urine sodium and chloride, whereas bartter syndrome has increased levels.[3] The management involves dehydration correction.

PBS usually presents in the neonatal age group.[4] Often, it presents before the typical manifestations of CF set in. In areas with low newborn screening, this metabolic feature coupled with failure to thrive can give a mammoth clue to physicians towards cystic fibrosis. This case report highlights the diagnostic dilemma that cystic fibrosis can be if its hallmark features have not yet presented.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent.

Conflicts of interest

There are no conflicts of interest.

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