

## A rare case report of barter syndrome type IV

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- Bartter syndrome is a rare inherited salt losing tubulopathy presenting as metabolic alkalosis with normotensive hyperreninemia and hyperaldosteronism. It is caused by mutations of the genes encoding protein that transport ions across renal cells in the thick ascending limb of nephron. Bartter syndrome can be divided into 5 sub types. Our case fits into Type IV Bartter syndrome with sensorineural deafness defect in the chloride channel. The disease is not curable. Disability depends on severity of mutation. Lifelong medication needed. Prognosis is generally good with early stabilization of metabolic abnormalities.
- We report here a case of two month old girl with Bartter syndrome with bilateral sensorineural deafness. More than hundred cases of Bartter syndrome were identified. This is the second case of Bartter syndrome with bilateral sensorineural deafness from the Indian community.
- Presenting features were failure to thrive, vomiting, polyuria and dehydration, vomiting, failure to gain weight, irritability, and constipation.
- h/opolyhydraminos 28th week of gestation .Normal vaginal delivery .cried immediately after birth . Neonatal period not significant. Breastfed every 2 hours now but baby still craves for feeds.
- On examination – lethargic, hypotonic, not responding to sound.
- Child exhibited metabolic alkalosis, ( pH7.6), hypokalemia(1.5 mEq/l), hypochloremia(49 mEq/l) and hyponatremia.(111 mEq/l).
- Disproportionate urinary wasting of sodium, potassium and chloride were seen.
- There is urinary loss of chlorine(127mmol/lit), hypercalciuria, serum aldosterone levels are high (600ng/lit),normal mg levels.
- USG abdomen showed nephrocalcinosis & BERA – profound bilateral sensoryneural hearing loss.
- Diagnosed as antenatal barter syndrome type IV with sesoryneural hearing loss.
- Child showed marked improvement with Ibuprofen and potassium supplementation on regular follow up for electrolyte estimation and SND.

**Key Message:** Suspect metabolic disorder in FTT. Early intervention improves the outcome.

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