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Pseudohypoaldosteronism type 1 in a Neonate Presenting with Acute kidney Injury, life-threatening Hyperkalemia and positive family history

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Back ground: Pseudohypoaldosteronism type 1 (PHA1) is a life-threatening disease that causes severe hyperkalemia with life threatening arrthymia resulting in cardiac arrest, acute kidney injury, if not treated immediately or if diagnosis is missed .

Objective: To report a case of a newborn with decreased intake of feeds and lethargy, finally diagnosed with Autosomal recessive pseudohypoaldosteronism type 1 with homozygous variant in exon 13 of the SCNN1A gene of alphaENAC(epithelial sodium channel) subunit.
In this article, we report a new born girl born third in order to consanguineous parents at 9 days of life with complaints of decreased intake of feeds, lethargy and yellowish discoloration of skin, eyes. She had a stormy clinical course with refractory hyperkalemia, salt wasting crisis and acute kidney injury requiring prolonged hospitalization. There was a family history of first sibling diagnosed with pure red cell aplasia followed by second sibling with sudden death on 7th day of life.

Results : Hyponatremia, hyperkalemia, severe metabolic acidosis, high creatinine despite adequate treatment were noted. Normal 17- OH-progesterone values, high Plasma Renin, Aldosterone and Cortisol values with absence of hyperpigmentation and no renal anomalies or adrenal hyperplasia on Ultrasonography. No response was seen with high dose of Fludrocortisone. This established diagnosis of PHA type 1.Refractory hyperkalemia got corrected initially only with peritoneal dialysis which recurred once dialysis was stopped with in few days despite continuing on other antihyperkalemic medications.Genetic analysis revealed a 1 with homozygous variant in exon 13 of the SCNN1A gene of alphaENAC subunit c.1727T>C which changes Leu576 in to Proline. Both parents and sibling were found to have a heterozygous variant in exon 13 of the SCNN1A gene of alphaENAC subunit c.1727T>C.

Conclusion : This may be probably the second case of PHA1 confirmed by genetic analysis in India and first in south India which highlights the role of consanguinity and severity of the disease.

Keywords : Psudohypoaldosteronism type 1, Hyperkalemia, Familial.