VIEWER'S CHOICE

Neonate With Fatal Genetic Disorder

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A preterm small for gestational age (SGA) male baby 2nd sibling of non consanguineous mating presented with convulsion, significant polyuria and weight loss.

Antenatal history: There was history of severe polyhydramnios at 26 weeks of gestation for which indomethacin treatment was required for 5 weeks. Indomethacin was discontinued at 31 weeks due to fetal right heart overload. There was recurrence of polyhydramnios within 2 weeks. Antenatally, structurally fetus was normal with no obvious cardiac, renal, skeletal or GIT anomalies. There were no hard or soft markers for chromosomal abnormalities. Diagnostic amniocentesis was done and fetal karyotyping (FISH analysis) showed normal signal pattern for chromosomes 18, 13, 21 and sex chromosomes. Amniotic fluid electrolytes were within normal limits.

Emergency caesarean section was preformed at 34weeks following Doppler flow changes of high resistance flow in umbilical artery, decrease in fetal breathing movements and non reassuring NST. The child delivered preterm, was normotensive with good APGAR with weight of 1.94kg.

Brief secondary apneic spell, excessive pulmonary alveolar fluid was noted for which the child needed endotracheal intubation and ventilation for fetal fluid. Neonate was ventilated for 14 hours. No hypoxia or acidosis was noted at anytime. Post extubation period was uneventful. Feeds were started on 3rd day of life. Child developed seizures on 3rd & 8th day due to hyponatremia. Also excessive weight loss (30%) and polyuria in the first three days of life were noted. Blood sugar and calcium, magnesium were normal. Symptomatic hyponatremia was corrected with appropriate replacement. Infant was hemodynamically stable at all times. Intracranial ultrasound, echocardiography and abdominal ultrasound were normal. A repeat ultrasound of abdomen after 14 days showed evolving nephrocalcinosis.

Serum cortisol level, 17OH progesterone, Uric acid were normal. Renin, aldosterone, prostaglandin levels were not done. Arterial blood gas showed metabolic alkalosis. There was a positive family history of paternal uncle being affected with similar illness. The child was treated with salt supplementation and oral indomethacin. The child was diagnosed as Neonatal Bartter's Syndrome (Seyberth hyperprostaglandin E syndrome)

Blood profile	**June 29**	**July 2**	**July 4**	**July 6**	**July 7**	**July 10**	**July 14**
Sodium (mEq/L)	**123**	123	132	**121**	127	129	130
Potassium (mEq/L)	5.9	4.5	4.5	5.4	4.8	**3.1**	4.9
Chloride (mEq/L)		100	103	100	101	102	
Bicarbonate (mEq/L)		18.9	23.4	25.4	21.2	23.8	25.4
Urea (mg%)	63		62	48	21.3		
Creatinine (mg%)	1.3		1.2	1.1	0.8		
B. Sugar (mg%)	61				58		
Calcium (mg%)	8.9						8.0
Magnesium		2.6					2.1
Ammonia (mmoL/l)		30					
Urinary Profile							
Urinary sodium (mEq/L)			94	66			44
Urinary chloride (mEq/L)			54		102		38
Urinary potassium (mEq/L)			10.6				22.4
Urine specific gravity	1.005						
Urinary 17 OH ketosteroids	**Normal**						
Urinary Creatinine (mg %)							20
Urinary Calcium (mg %)			11.6				17
Urine calcium/creatinine Ratio							**0.85**

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