CASE REPORT

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Central Diabetes Insipidus Association with Rare Chromosomal Anomaly Trisomy 9p and Monosomy 18q

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This case report was presented as a poster at 28th Annual Meeting of The European Society of Paediatric and Neonatal Intensive Care, June 6-9 2017, Lisbon, Portugal.

ABSTRACT Term infant was admitted to Neonatal Unit on day one due to dysmorphic facial features and poor feeding. Serum electrolytes were checked at 24 hours of life and showed serum sodium of 143mmol/L and normal renal function test. Serum sodium rose to 148mmol/L at 48 hours and 155 mmol/L at 72 hours of life. On day three; high serum osmolality, low urine specific gravity: 1.005, low osmolality 105 mOsm/kg. Due to suspicion of Diabetes insipidus (DI) one dose of subcutaneous desmopressin (DDAVP 100ng) corrected his urine specific gravity, serum sodium, and serum osmolality to within normal range (signify central DI). Chromosomal analysis showed translocation of chromosomes 9p and 18q.

Keywords: Trisomy 9p & monosomy 18p; central diabetes insipidus; Desmopressin (DDAVP); serum osmolality

A term male infant born to 38 years old G3P1^{+2} female via Cesarean section due to breech presentation. In current pregnancy she has additional risk factors of gestation diabetes, advanced maternal age and polyhydromnios on antenatal ultrasounds.

Infant was admitted to Neonatal Unit on day one due to dysmorphic facial features and poor feeding. Serum electrolytes were checked at 24 hours of life and showed serum sodium of 143 mmol/L, BUN of 3.6 mmol/L, and creatinine of 68 umol/L. Serum sodium raised to 148 mmol/L at 48 hours and 155 mmol/L at 72 hours of life. So suspicion of diabetes insipidus (DI) was raised. On day three, it was also noted that serum glucose (2.7mmol/L [3.9-7.8]), serum osmolality: 320mOsm/kg [282-300], urine specific gravity: 1.005, urine osmolality of 105 mOsm/kg and urine sodium of 86mmol/L.

On subsequent days further radiological investigations were conducted. Renal ultrasound was reported as normal. Cranial Ultrasound showed enlarged lateral ventricles, measuring 5.2 mm x 3.3 mm.

Computed Tomography (CT) Brain: prominent lateral ventricles, mild trans ependymal edema, normal 3rd and 4th ventricles, otherwise normal findings.

We determined that the cause of the patient's hypernatremia was likely secondary to DI. Endocrine team was contacted and conducted DDAVP [a

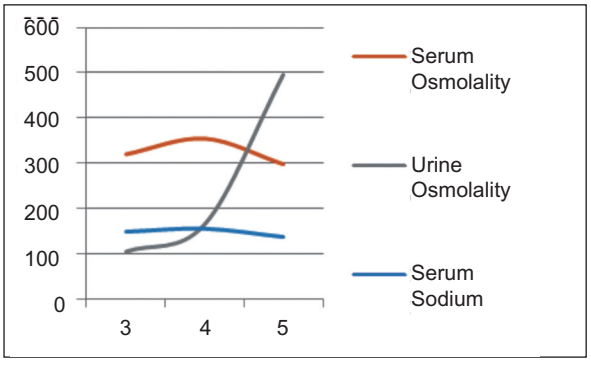

FIGURE 1: On day five: (DDAVP Received on day 4).

trade name of desmopressin, 1-deamino-8-D-arginine vasopressin] challenge test on day four, to determine if the DI was central or nephrogenic.

One dose of subcutaneous (SC) desmopressin, (DDAVP 100 ng) corrected his urine specific gravity, serum sodium, and serum osmolality to within normal range.

Thus, we concluded that his ADH receptors at the level of the nephron were functioning properly, and his DI was central in etiology. The infant would require synthetic ADH for the rest of his life.

So, on day five (DDAV received on day 4); Serum sodium 136 mmol/L, serum osmolality: 298 mmol/kg, urine osmolality: 493 mOsm/kg, urine sodium: 35 mmol/L, FSH: <0.5 (0.8-9.0) and LH: <0.5 (0.8-7.6) (Figure 1).

The low levels of other pituitary hormones leads us to believe the infant has perhaps an underlying pan-hypo- pituitarism.

Results of karyotyping from day one showed a translocation of chromosomes 9p and 18q.

This chromosomal abnormality has never been documanted before.

Since chromosome 9 is half of 18, it is possible that this is an imbalance of chromosomes rather than a true translocation.

PHENOTYPIC PERTINENT PHYSICAL EXAM FINDING

- Birth weight of 2.92 kg (9th Percentile, Length 50 cm (50th Percentile), HC 34 cm (25th Percentile); on WHO-UK/Ireland growth Cenile chart for boys).

EAR ANOMALY

- Low set ears with abnormal antihelices.
- Sensory-neural hearing loss.

HEAD AND NECK

- Widely open sagittal suture with large communicating anterior and posterior open Fontanelle (Anterior Fontanelle 4 x 5 cm/Posterior Fontanelle 3 x 4 cm)
- Micrognathia,
- Sclera icteric
- Short neck with thick nuchal pad

GENITALIA

- Male with undescended testes bilaterally

EXTREMITIES

- Contractures at proximal interphalangeal joints bilaterally,
- Clinodactyly,
- Short femurs, and small feet.

DISCUSSION

Diabetes derived from a Greek word "*diabainein*" meaning Siphon-to pass through; referring to excessive urination associated with the disease.

Insipidus derived from the Latin word meaning "without taste," DI involves the passing of tasteless urine due to low sodium content.

Mellitus is also a Latin word meaning "sweet." Due to the excessive sugar found in blood and urine of a diabetic patient.[1]

DI is a life threatening condition, as it causes fluid and electrolyte imbalance that results in polyuria, weight loss, diluted urine, severe hypernatremic dehydration.[2]

There are a variety of pathologic processes can impede the production or secretion of adrenal diuretic hormone (ADH) and leads to Central DI, which includes idiopathic, infectious, trauma, malignancy, genetic, familial, intracranial haemorrhage and hypoxia.[3]

Synthetic ADH analogue, DDAVP is the main stay of treatment in central DI. It is available in intranasal, oral, SC, and intravenous preparation.

Polyuria in central DI could also be control with a paradoxical antidiuretic effect of thiazide diuretics.[4,5]

Genetic counselling is necessary, because the recurrence risk significantly increases if one of the parents is a carrier. Antenatal diagnosis is possible by conducting amniocentesis, chorionic villus sampling and cytogenetic testing including FISH. The details about this rare genetic condition could be taken from the "Unique understanding of chromosomal disorders" website for support and information.[6,7] http://www.rarechromo.org/html/Disorder Guides.asp and NORD (National Organisation for Rare Disorders) 18-monosomy-18p/.

Informed Consent

Consent was given by the parents to publish these results. Karyotype report from OLCHC, Genetics lab (Ireland).

Source of Finance

During this study, no financial or spiritual support was received neither from any pharmaceutical company that has a direct connection with the research subject, nor from a company that provides or produces medical instruments and materials which may negatively affect the evaluation process of this study.

Conflict of Interest

No conflicts of interest between the authors and / or family members of the scientific and medical committee members or members of the potential conflicts of interest, counseling, expertise, working conditions, share holding and similar situations in any firm.

Authorship Contributions

All authors contributed equally while this study preparing.

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