CONGENITAL NEPHROGENIC DIABETES INSIPIDUS IN PREGNANCY

CASE REPORT

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INTRODUCTION

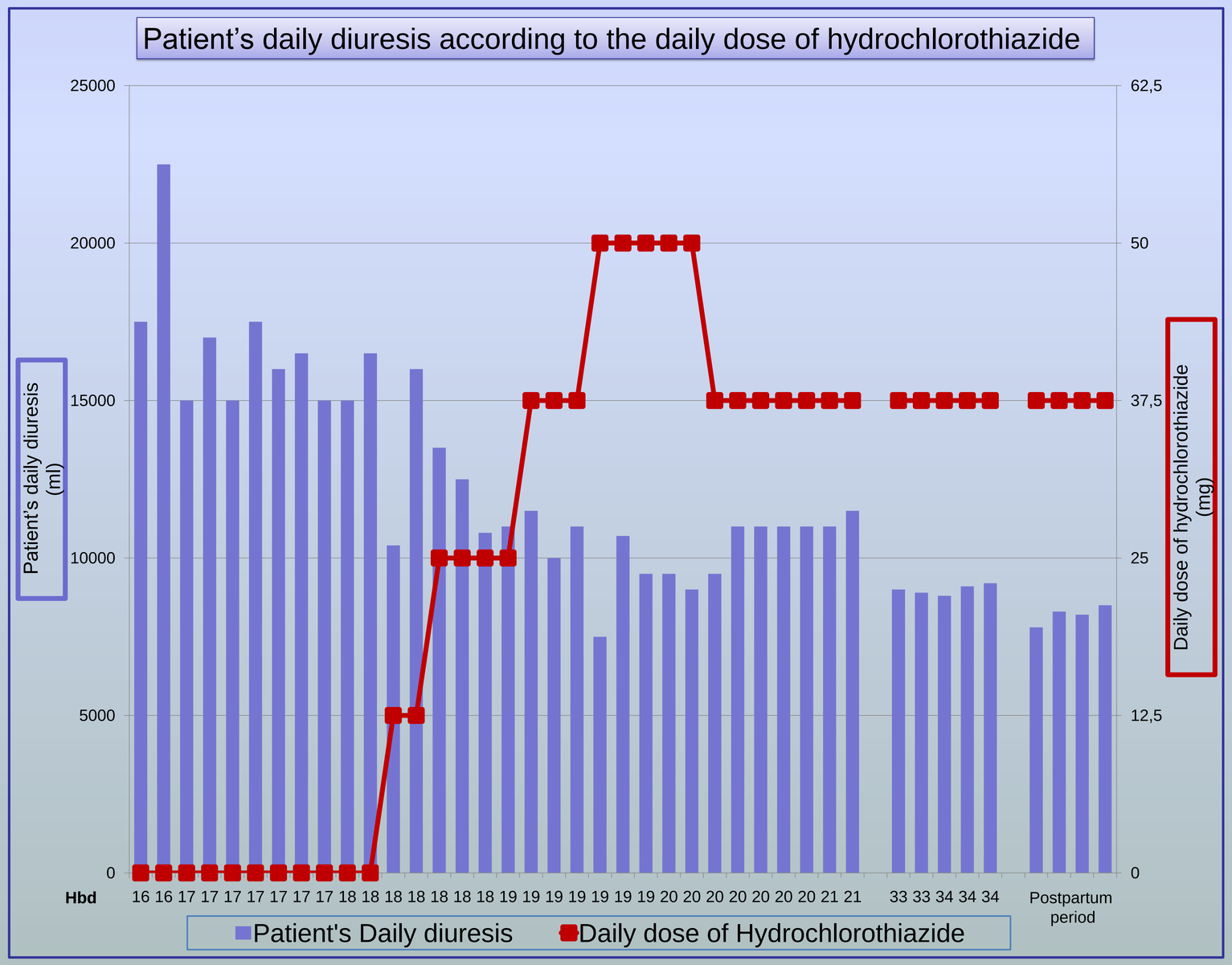
Congenital nephrogenic diabetes insipidus (NDI) is a hereditary renal disorder characterized by failure to concentrate urine in response to antidiuretic hormone (ADH). 90% of cases are caused by X-linked mutations of the ADH V2 receptor and 10% by autosomal mutations of the aquaporine 2 water channel (AQP2). The prevalence for NDI caused by AQP2 mutations is about 1/20mln births. No data was found about an overall prevalence at females and their chances for a successful pregnancy. We present a case of woman patient diagnosed with NDI in her childhood and the effect of pregnancy on the disease outcome.

CASE DESCRIPTION

A 23-year-old primigravida at 15 weeks of gestation was referred to the hospital with dysuria and abdominal pain. The patient had polydipsia and polyuria, at admission her urine volume reached 17L/day. Anamnesis revealed diagnosis of NDI in her childhood and hydrochlorotihiazide treatment until the age of 18. Her twin sister died at the age of 6 because of severe dehydration. Other family members asymptomatic. Patient claimed ingesting big quantities of fluid for as long as she remembers, during her adulthood the symptoms didn’t impact on the quality of her life. In her early pregnancy she observed gradual aggravation of the symptoms: an excessive thirst (up to 18L/day), increased urine output. Upon examination the patient without definite evidence of dehydration, blood pressure 101/80 mmHg. The serum sodium 132 mmol/L, urine specific gravity 1,003, serum creatinine 0,5mg/dl. Plasma and urine osmolality: 276mOsm/L and 78mOsm/L respectively, culture urine positive. USG detected bilateral pelvicalyceal dilatation and enlarged bladder.

INTERVENTION & RESULTS

The treatment strategy aim was to reduce the diuresis and urine retention, prevent recurrent urinary infections and thus lower the risk of miscarriage or preterm delivery. The patient received hydrochlorothiazide in doses up to 50 mg/day which reduced the diuresis to 7L/day. Electrolytes supplementation (K, Mg) provided under their levels control in serum and 24-h urine collection. During therapy patient developed gestational diabetes, diet-controlled. Regular monitoring of the fetus provided. The optimal dose of thiazide determined to be 37,5mg/day. Elective C-section at 38th week of gestation, female neonate delivered, Apgar score 10. The woman with infant discharged from hospital in the 8th day of neonates life. The results of genetic testing in development, as there has been yet identified one autosomal recessive mutation of AQP2 in patient and her mother. To determine if it’s homozygous or compound heterozygous the rest of the family must be tested (especially the patient’s father). Additionally, the same treatment strategy performed during the 2nd pregnancy resulted in another successful delivery.



CONCLUSIONS

Careful monitoring of the patient’s fluid balance is essential as the outcome of NDI tends to aggravate in pregnancy. Although thiazide diuretics belong to FDA pregnancy B category (the risks of fetus’ thrombocytopenia, haemolytic anaemia, electrolyte imbalances), during NDI manifestations, they can decrease the diuresis - in the presented case the benefits outweigh the potential risk.

Key words: diuretics, nephrogenic diabetes insipidus, pregnancy, hereditary, AQP2 water channel

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