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Cerebral Salt Wasting and Fludrocortisone in a Child with Lissencephaly, Holoprosencephaly, Cleft lip and Palate

Se Jin Park¹, Ki Soo Pai², Jae Il Shin³

¹Department of Pediatrics-Nephrology, Geje Children's Hospital, Korea, Republic of

²Department of Pediatrics-Nephrology, Ajou University Hospital, Korea, Republic of

³Department of Pediatrics-Nephrology, Severance Hospital, Korea, Republic of

Case Study: Lissencephaly, which literally means smooth brain, is a rare brain-formation disorder caused by defective neuronal migration during the 12th to 24th weeks of gestation, resulting in a lack of development of brain folds and grooves. Symptoms of the disorder include significant developmental delay, infantile spasm, epilepsy, and muscle spasticity or hypotonia. Although life expectancy was said to be shortened in the past, with advances in therapy, most children will be able to survive longer than two years of age.

Holoprosencephaly is another brain-formation disorder that prosencephalon (forebrain of the embryo) fails to develop into two hemisphere. There are three classifications of holoprosencephaly: Alobar, in which the brain has not divided at all, is usually associated with severe facial deformities. Semilobar, in which the brain's hemispheres have somewhat divided, causes an intermediate form of the disorder. Lobar, in which there is considerable evidence of separate brain hemisphere, is the least severe form.

Affected children may display unusual facial appearance such as cleft lip and palate, cyclopia, mental retardation, and electrolyte imbalance. In the electrolyte disturbances, differential diagnosis of hyponatremia requires a systematic and sequential approach due to the different treatment between the syndrome of inappropriate antidiuretic hormone secretion marked by inappropriate retention of free water and cerebral salt wasting (CSW) characterized by excessive urinary loss of sodium and resulted in polyuria and extracellular volume contraction. We herein present a 9-year-old child with CSW and successful treatment with fludrocortisone in the above brain-formation disorders.