Case Report

A Case of Galloway-Mowat Syndrome with Classic Clinical Triad in the Neonatal Period

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Galloway-Mowat syndrome (GMS) is a rare autosomal recessive disorder comprising of early-onset nephrotic syndrome and central nervous system involvement including microcephaly, seizure and developmental delay. Although hiatal hernia is no longer considered essential findings for diagnosis, clinical triad of GMS included nephrotic syndrome, neurological manifestations, and hiatal hernia in the original description. We experienced a case of newborn with GMS presenting these clinical triad in neonatal period. A male infant weighing 2,250 g was born at gestational week 39 +3 by cesarean section. The patient revealed mild dysmorphic facial features and microcephaly. On day 7, Nissen fundoplication was done because of hiatal hernia with gastric volvulus. At the age of 2 weeks he developed nephrotic syndrome with proteinuria and hypoalbuminemia. This is the first case of GMS that three classic findings were present in neonatal period in Korea.

Key Words : Galloway-Mowat syndrome, Nephrotic syndrome, Newborn

Galloway-Mowat syndrome (GMS) is a rare malformation syndrome characterized by intrauterine growth retardation (IUGR), early-onset nephrotic syndrome, hiatal hernias and central nervous system involvement such as microcephaly, cerebellar atrophy and developmental delay. In the original description, GMS was reported as a clinical triad of nephrotic syndrome, neurological manifestations, and hiatal hernia. However, hiatal hernia is no longer considered essential findings for diagnosis. In our knowledge, the patients with classic clinical triad in neonatal period have not been reported in Korean population. Therefore, the authors herein describe an infant showing hiatal hernia, neurologic abnormalities such as microcephaly and nephrotic syndrome in neonatal period.

A 31-year-old woman was referred to our hospital at 39 weeks of gestation because of congenital diaphragmatic hernia and intrauterine growth restriction, which were suspected by ultrasonography at 36 weeks of gestation. A male infant weighing 2,250 g was born at gestational week 39 +3 by cesarean section. Head circumference was 29 cm (below 3 percentile). The patient was the first baby of nonconsanguineous parents. There was no family history of congenital anomaly. Apgar scores were 5 and 7 at 1 and 5 min, respectively. Physical examination revealed mild dysmorphic facial features such as high nasal bridge, micrognathia and narrow forehead.
Chest radiography and upper gastrointestinal series revealed hiatal hernia with gastric volvulus (Fig. 1). On day 7, Nissen fundoplication was done. A feeding problem was apparent and a nasogastric tube was required for nutrition. At the age of 2 weeks, he became edematous and developed nephrotic syndrome with proteinuria and hypoalbuminemia. The 24 hour excretion of urinary protein was 2.871 g. Fundus in optic evaluation was normal without microcoria. On day 28, open renal biopsy was done. Renal biopsy showed focal segmental glomerulosclerosis (Fig. 2). Magnetic resonance imaging (MRI) of brain was suggestive of lissencephaly (Fig. 3). The patient’s karyotype indicated 46, XY. Screening for metabolic disease was unremarkable. The patient was discharged while keeping tubal feeding at 43 days of age.

The patient was readmitted because of afebrile convulsion at 4 month of age. The interictal electroencephalogram during sleep was moderately abnor-