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Case Report

Galloway-Mowat syndrome: Prenatal ultrasound and perinatal magnetic resonance imaging findings

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Abstract

Objective: To present prenatal ultrasound and perinatal magnetic resonance imaging (MRI) findings of Galloway-Mowat syndrome.
Case Report: A 31-year-old woman, gravida 3, para 2, was referred for genetic counseling at 29 weeks of gestation because of abnormal ultrasound findings and a previous child with Galloway-Mowat syndrome. During this pregnancy, microcephaly, intrauterine growth restriction (IUGR), and oligohydramnios were first noted at 27 weeks of gestation. Repeated ultrasounds showed microcephaly, IUGR, and oligohydramnios. MRI performed at 32 weeks of gestation showed reduced sulcation of the brain, pachygyria, poor myelination of the white matter, and cerebellar atrophy. A diagnosis of recurrent Galloway-Mowat syndrome was made. At 40 weeks of gestation, a 2,496-g female baby was delivered with microcephaly, a narrow slopping forehead, epicanthic folds, microphthalmos, a highly arched palate, a small midface, a beaked nose, thin lips, large low-set floppy ears, clenched hands, and arachnodactyly. Postnatal MRI findings were consistent with the prenatal diagnosis. Renal ultrasound showed enlarged bilateral kidneys with increased echogenicity. At the age of 2 weeks, the infant became edematous and developed nephrotic syndrome.
Conclusion: Microcephaly, IUGR, and oligohydramnios are significant ultrasound triad of fetal Galloway-Mowat syndrome. Prenatal ultrasound diagnosis of microcephaly, IUGR, and oligohydramnios in late second trimester or in early third trimester should alert clinicians to the possibility of Galloway-Mowat syndrome and prompt a detailed search of abnormal sulcation, cortical gyral maldevelopment, and cerebellar atrophy by fetal ultrafast MRI.

Keywords: Cerebellar atrophy; Galloway-Mowat syndrome; Magnetic resonance imaging; Prenatal diagnosis; Ultrasound

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Introduction

Galloway-Mowat syndrome (online Mendelian inheritance in man 251300), or microcephaly, hiatus hernia and nephrotic syndrome, or nephrosis–neuronal dysmigration syndrome, or nephrosis–microcephaly syndrome, is a rare autosomal recessive disorder. This syndrome was first described by Galloway and Mowat in 1968 [1] in a brother and a sister with microcephaly, hiatal hernia, and nephrotic syndrome. Patients with Galloway-Mowat syndrome will develop early-onset intractable seizures and nephrotic syndrome, psychomotor delay, mental retardation, and death in early childhood because of renal failure [2]. Galloway-Mowat syndrome is characterized by microcephaly; early-onset corticosteroid-resistant nephrotic syndrome; developmental delay; and central nervous system abnormalities, such as defects of neuronal migration, hypomyelination, and cerebellar atrophy [3]. Other additional findings include a slopping forehead, flat occiput, hypertelorism, ptosis, corneal opacity, cataract, microphthalmos, hypoplastic iris, optic atrophy, large low-set floppy ears, a small midface, a beaked nose, high-arched palate, micrognathia, club feet, camptodactyly, flexion contractures of joints, hypoplastic nails, hiatal hernia, eventration of diaphragm, ovarian agenesis, atrophic thymus, calcification of the intervertebral discs, platybasia, Dandy-Walker malformation, aqueductal stenosis with hydrocephalus, and central canal malformation [3]. Arachnodactyly has been observed in some Taiwanese patients affected with Galloway-Mowat syndrome [4–6]. Here, we present prenatal ultrasound and perinatal magnetic resonance imaging (MRI) findings of recurrent Galloway-Mowat syndrome in a fetus.

Case report

A 31-year-old woman, gravida 3, para 2, was referred for genetic counseling at 29 weeks of gestation because of

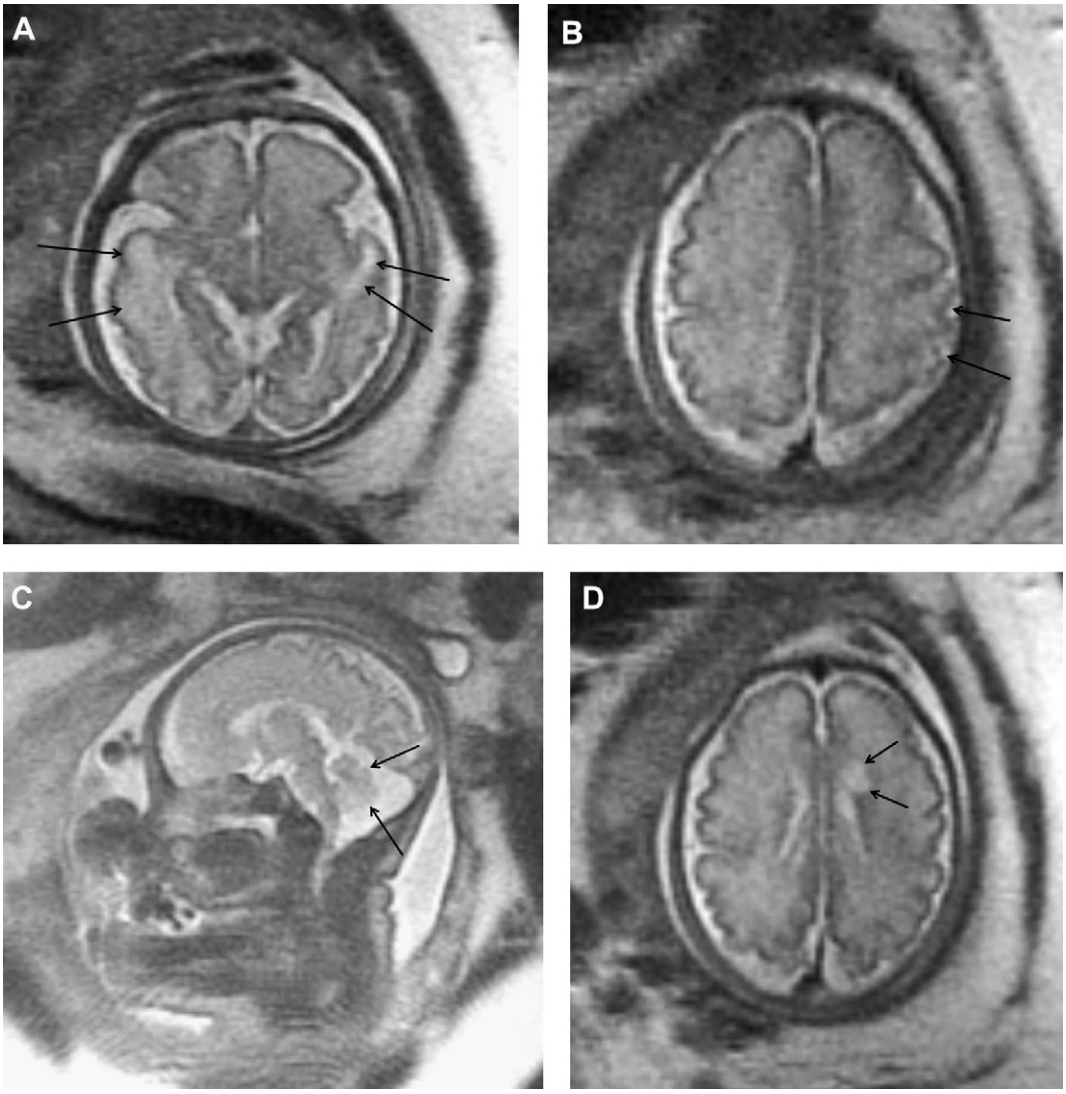

Fig. 1. Fetal ultrafast magnetic resonance imaging at 32 weeks of gestation shows (A) hypomyelination with increased T2 signal intensity over bilateral temporal white matter (arrows); (B) pachygyria with effacement of cortical gyri (arrows); (C) cerebellar atrophy (arrows); and (D) a cyst arising from the frontal horn of left lateral ventricle (arrows).