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LETTER TO THE EDITOR

Prenatal Ultrasound and Magnetic Resonance Imaging Findings of Fetal Akinesia Deformation Sequence with Multiple Pterygium Syndrome

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A woman aged 27 years, gravida 2, para 1, was referred for genetic counseling at 27 weeks of gestation because of reduced fetal movement, scoliosis, and clubfoot detected by ultrasonography. She and her husband were non-consanguineous and healthy and had a son aged 3 years. The woman did not abuse drugs, and she did not have any metabolic conditions or neuromuscular disorders, such as myasthenia gravis, during this pregnancy. Amniocentesis revealed a karyotype of 46,XY, and array comparative genomic hybridization analysis revealed no genomic imbalance. A level 2 ultrasound at 29 weeks of gestation revealed a singleton male fetus with fetal biometry equivalent to 29 weeks, reduced fetal movement, severe kyphoscoliosis, pulmonary hypoplasia, and arthrogryposis (Fig. 1). Magnetic resonance imaging (MRI) study additionally showed multiple pterygia (Fig. 2). The fetus had intrauterine fetal death, and a 1234-g fetus was delivered with down-slanting palpebral fissures, hypertelorism, a depressed nasal bridge, low-set ears, micrognathia, joint contractures, and multiple pterygia (Fig. 3). Radiographic studies showed kyphoscoliosis and a restrictive left chest (Fig. 4). Molecular analysis revealed no mutation in the genes of *CHRNA1*, *CHRND*, *CHRNG*, *RAPSN*, or *DOK7*.

Fetal akinesia deformation sequence (FADS) is a clinically and genetically heterogeneous disorder that is characterized by fetal akinesia, arthrogryposis, intrauterine growth restriction, congenital abnormalities, such as cystic hygroma, pulmonary hypoplasia, facial cleft, cardiac defects, cryptorchidism, and intestinal malrotation, and occasionally pterygia of the limbs [1]. The incidence of FADS is about 1:15,000 births [2]. FADS may phenotypically overlap with the lethal type of multiple pterygium syndrome (LMPS). FADS/LMPS can be caused by neuropathy, muscular disorders, neuromuscular junction disorders, myasthenia

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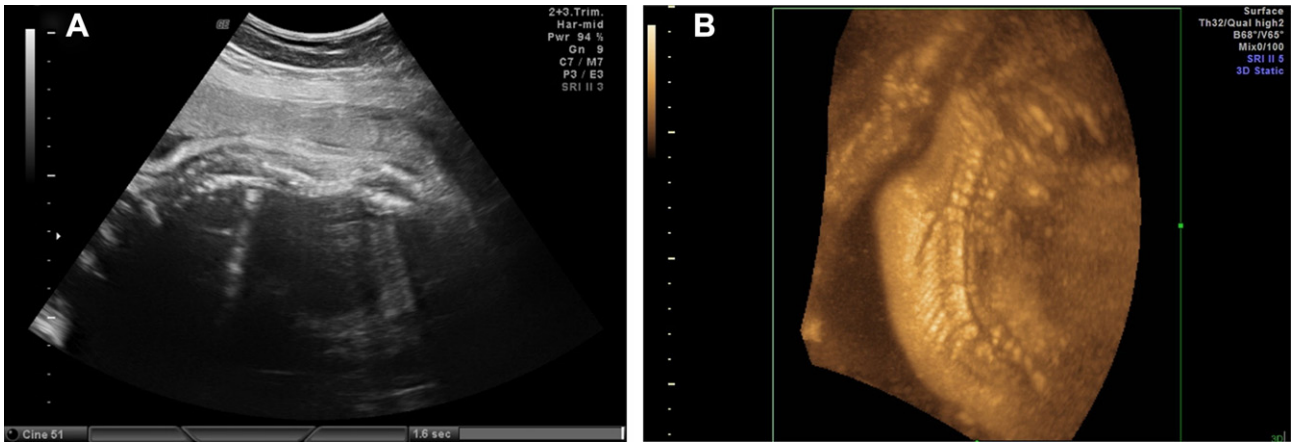

Fig. 1 Prenatal (A) two-dimensional; and (B) three-dimensional ultrasound demonstration of kyphoscoliosis.

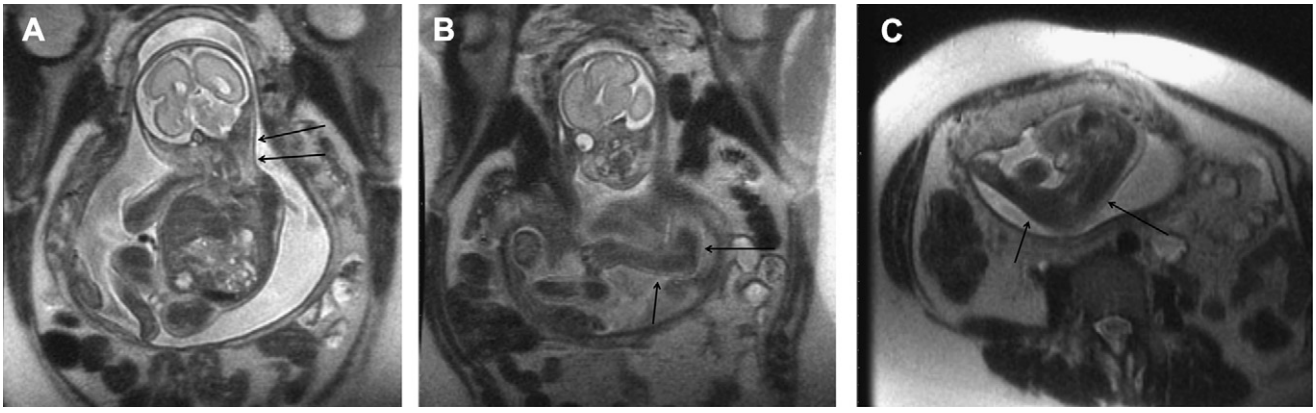

Fig. 2 Magnetic resonance imaging of the fetus shows (A) webbing of the neck (arrows); (B) pterygia with fixed deformity of the elbow (arrow); (C) pterygia with fixed deformity of the left lower limb (arrow).

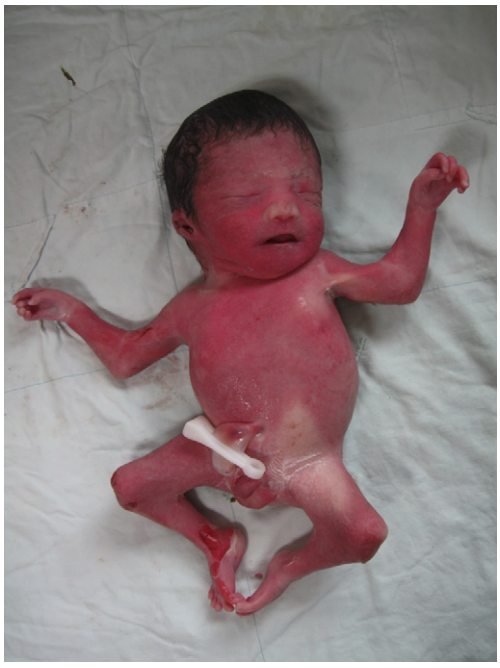

Fig. 3 Fetus at birth.

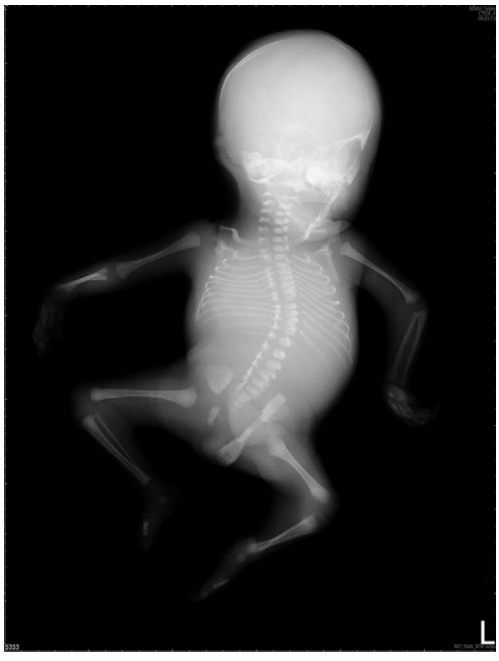

Fig. 4 Radiography shows severe kyphoscoliosis.

gravis, restrictive dermopathy, in utero restriction space of the fetus, vascular compromise, ischemia, teratogenic exposures, circulating maternal antibodies to neurotransmitters, myelin, and muscle proteins [1,3,4]. Prenatal ultrasonography findings of FADS/LMPS include lack of extremity motions, persistent abnormal limb posture, polyhydramnios, pulmonary hypoplasia, a short umbilical cord, increased nuchal translucency, cystic hygroma, and hydrops fetalis [5,6]. Fetal MRI has been suggested as a useful adjunct to ultrasound in evaluating associated central nervous system abnormalities [6,7]. Prenatal diagnosis of FADS/LMPS should include a differential diagnosis of spina bifida, trisomy 18, fetal constraint, body stalk anomaly, caudal regression sequence, fetal hypoxia, amniotic band sequence, fetal neck masses, joint and vertebral anomalies, and iniencephaly [8].

Acknowledgments

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