A 40 years-old patient, G4P3A1, was initially referred to our obstetrical high-risk center during her first pregnancy for an abnormal morphologic ultrasound at 21 weeks. She was exposed to a severe carbon monoxide poisoning at 19 weeks of pregnancy that required hyperbaric oxygen therapy. Her 21st week ultrasound showed an asymmetric ventriculomegaly and a corpus callosum agenesis. The fetal magnetic resonance imaging (MRI) showed extensive cerebral clastic lesions, hypoplasia of the corpus callosum and delayed gyration. She had a vaginal delivery at term and her young boy of 8 years old has an intellectual disability, significant spasticity, a language disorder, strabismus and optic atrophy. Her second pregnancy was normal and she delivered of a healthy boy at term.

At her third pregnancy, the morphologic ultrasound at 20 weeks showed the same fetal brain abnormalities than the first pregnancy as bilateral ventriculomegaly, suspicion of corpus callosum agenesis. This male fetus had bilateral adductus thumbs. The fetal MRI found a triventricular dilatation, an important partial agenesis of the corpus callosum, an atrophic protuberance and an atrophic cerebral parenchyma. Given an evocative diagnosis of L1CAM, the first child was investigated for the mutation which was positive. The patient was found to be L1CAM carrier and the mutation was found in the fetus. She had a cesarean section at term because of breech presentation.

L1CAM syndrome is a mild to severe X-linked developmental congenital disorder characterized by hydrocephalus of various severity degrees, intellectual deficit, lower limb spasticity, and adducted thumbs. We report antenatal diagnosis of L1CAM in a family because of recurrence of fetal brain abnormalities. Both babies are followed in pediatric neurology.