In Utero Diagnosis of Niemann-Pick Type C in the Absence of Family History

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Résumé en anglais	Niemann-Pick type C (NPC) disease is a recessive disorder that results in unesterified cholesterol accumulating in the lysosomal and late endosomal system. It is caused by mutations in NPC1 or NPC2 genes and leads to systemic and neurodegenerative symptoms. Few cases of prenatal presentation of NPC have been reported and only two cases in the absence of previous family history, indicating the diagnosis is particularly difficult in such a situation. We report a prenatal diagnosis of NPC in a couple without family history. An ultrasound screening at 22 weeks of gestation (WG) detected fetal ascites and hepatomegaly, which were still present at 25, 27, and 29 WG, and a splenomegaly progressively appeared. No placentomegaly or other signs of hydrops fetalis were observed. The diagnostic of NPC was prenatally confirmed by a filipin test and NPC1 sequencing and multiplex ligation-dependent probe amplification assay which revealed a maternal missense mutation (c.2608T>C; p.Ser870Pro) and a paternal deletion of exons 5 to 25. This additional prenatal case of NPC suggests that even in the absence of family history, fetal ascites associated with splenomegaly but no hydrops should nonetheless arouse suspicion concerning this disease as a possible diagnosis.
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