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NIEMANN-PICK DISEASE TYPE C IN PATIENTS ENZYMATICALLY PROVEN AND NOVEL MUTATIONS DEFINED THE FIRST REPORT FROM LIBYA AND REVIEW OF LITERATURE

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Niemann-Pick disease type C (NP-C) is a rare neurovisceral disease, lipid storage disorder, is characterized by intracellular deposition of sphingomyelin in various body tissues. NPC is autosomal-recessive disease caused by mutations in *NPC1* and *NPC2* genes. Symptoms are variable and can occur at any age. In this study, we report on the first five patients with NP-C from Libya, the severity and outcomes were quite distinctive. NP-C in patient 1, manifested as prolonged neonatal jaundice and hepatomegaly during the first year of life, while dystonia, dysarthria and dysphagia were the warning signs for late diagnosis at age of 4 years. She unfortunately experienced respiratory and dermatological complications, and died at eight years old. In contrast, patient 2, hospitalized at age of 50 days as progressive hepatosplenomegaly and severe neonatal cholestatic hepatopathy, he succumbed to his disease at age of 3 months. Both patients 3 and 4 experienced liver dysfunction with prolonged neonatal jaundice. They recovered and have continued to achieve age-appropriate development to date. Patient 5, whose diagnosis is based on sibling screening, is asymptomatic. Upon analysis, lyso-SM-509 biomarker for the diagnosis in four patients had shown pathologically increased levels. *NPC1* mutational analysis revealed in patient 2 is homozygotic for a previously unreported homozygous duplication encompassing exons 10 and 11 of the *NPC1* gene. Patient 3 and 5 are sisters, are homozygotic for a previously unreported mutation in exon 13 of the *NPC1* gene, c.2027G>T(p.Ser676Ile); In Patient 4, detected a novel mutation multiple copies of exon 10 and 11 of the *NPC1* gene. The NP-C Suspicion Index (SI) is a screening tool offer considerable help in the diagnostic process of patients with suspicion of NP-C. Miglustat is a disease-specific treatment, for a favorable outcome, should be started immediately in patient with any type of neurological manifestation.

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