

JOINT EVENT

8th World Congress on

Rare Diseases & Orphan Drugs

and

Clinical Trials & Regulatory Affairs

November 21-22, 2018 | Paris, France

POSTER

8th World Congress on Rare Diseases and Orphan Drugs & Clinical Trials & Regulatory Affairs

November 21-22, 2018 | Paris, France



Djamel Eddine Ouail

Abderrahmane Mira University, Algeria

CO-Authors

**Fayçal Bouali, Djamil Si Ahmed and
Meriem Tebbani**

Abderrahmane Mira University, Algeria

Niemann Pick disease, about three cases

Niemann-Pick type B disease (NPD type B) is a Lysosomal disease with autosomal recessive inheritance, which is defined by Sphingomyelin accumulation within macrophages secondary to a deficiency of sphingomyelinase activity. We describe three adult cases of NPD type B.

Observations: patient CC born in 1999 the last of a brotherhood of 9 brothers from a non-consanguineous marriage, consults for type V splenomegaly evolving gradually from a young age without complications until then, the interview is poor apart from a notion of hematemesis of low abundance at the time of menstruation. Clinical examination is normal apart from splenomegaly. The blood test is normal. The enzymatic assessment found a weak activity of Sphingomyelinase acid, the genetic study had detected two homozygous mutations confirming the illness of Niemann-Pick. At the time of the family investigation the brother C A presented a Splenomegaly is the enzymatic assessment as well as the genetic study had confirmed the diagnosis.

Patient S A Born in 1982, married and mother of two children living in good health, consulted for febrile Polyarthralgia associated with headache. In his antecedents noted a thrombocytopenia of autoimmune allure followed in hematology and put under corticoids. The clinical examination found a Hepatosplenomegaly without portal hypertension, the biology revealed thrombocytopenia at 40 000 / mm³ without hemorrhagic syndrome, the rest of the balance sheet is without abnormality. The enzymatic assay and the genetic study confirmed the diagnosis.

The three patients benefited from the only symptomatic treatment while waiting for a specific treatment.

Biography

Djamel Eddine Ouail has completed his PhD from Algeirs University. He is Hospitalo University Assistant in the department of internal medicine at University Hospital Center of Bejaia, Abderrahmane Mira University of Bejaia Algeria.

Email id: djameleddineouail@gmail.com

Notes: