Discussion
Muscle biopsy confirmed respiratory chain disorder with complex magnetic resonance showed global loss of volume of white matter. 1-antitrypsin deficiency, increased lactic acid and T4 low. Cerebral cal as clinical.

Results
Introduction
If the patient showed acquired microcephaly and absent speech associated with convulsions and angelman-like features, the most probably diagnosed is Angelman syndrome. The screening for mutations in chromosome 15 diagnosed the syndrome. If the patient has also behavioral disturbances with family history of learning disabilities, determination of urine creatine is obligatory to exclude creatine transport defect. If all these tests are negative and the patient has unrelated organs involved, we need to exclude respiratory chain disorder and muscle biopsy is mandatory.

Conclusion
We emphasize the importance of studying more of cases (Clinic - Genetic) to put an update on the current classification.

The early therapeutic in the management of GD is still advantageous.

Clinical and Therapeutic Aspects of Gaucher’s Disease in Children
A Benketira. Pediatric, Military Hospital Regional University of Oran, Oran, Algeria

Introduction
Gaucher’s disease is the most common of the inherited metabolic disorder known as lipid storage diseases. It is a lysosomal disease, autosomal recessive. It is caused by a deficiency of beta-glucocerebrosidase. The result is a substance called glucocerebroside to build up in cells of the body (Spleen, liver, lungs, bones and sometimes in the brain).

There are three clinical types:
- **Type 1**
  - 95%
  - 1/50000
- **Subacute**
  - Infants/Children
  - Doesn’t involve the brain
- **Type 2**
  - 1%
  - 1/150000
- **Acute/Deadly**
  - Newborn-06 months
- **Severe brain damage**
  - Type 3;
  - 5%
  - 1/100000
- **Chronic**
  - Juvenile/Adult

Brain-Liver-Spleen involvement appear gradually

Materials and Methods
It’s a baby 13 months old. He had hepatosplenomegaly with cytopenia. He had the neurological signs such pyramidal syndrome with contra version ocular without flutter.

The explanation concluded for the GD by the enzymatic dosage.

Results
After six years of follow up, enzyme replacement therapy (Imiglucerase) has demonstrated its effectiveness as well as biological as clinical.

Our observation has been raised the possibility of signs of brain involvement in the type 1.

The finding joins a few cases in the literature.

This data calls into question the traditional classification cited from above.

A155