Case 2 - Five-year-old boy, has acquired microcephaly since 12 months of age, absent speech, mental retardation, behavioral problems. Creatine Transport Defect was confirmed with high levels of creatine in urine and hemoglycid myosin mutation seen in the SLC6A8 gene.

Case 3 - Eleven-months-old boy, has acquired microcephaly since six months of age, absent speech, hypotonia. He also has α-1-antitrypsin deficiency, increased lactic acid and T4 low. Cerebral magnetic resonance showed global loss of volume of white matter. Muscle biopsy confirmed respiratory chain disorder with complex 2 deficiency - 25%.

Discussion 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 mutation in chromosome 15 diagnosed the syndrome. If the patient has also behavior 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.