P-096 - CRIM-NEGATIVE CLASSIC INFANTILE POMPE DISEASE PATIENT ON ERT SINCE 40 DAYS OF AGE

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\textbf{INTRODUCTION:} Pompe disease (PD) is a progressive and often fatal muscular disease caused by a deficiency of the lysosomal enzyme acid-glucosidase (GAA). It is a rare autosomal recessive disorder. The clinical spectrum of PD varies with respect to age at onset, rate of disease progression, and extent of organ involvement. The classic infantile form exhibit rapidly progressivity, prominent cardiomegaly, hepatomegaly, weakness and hypotonia. Death occurs due to cardiorespiratory failure in the first year. This form of PD represents the most severe end of the disease spectrum. The advent of specific enzyme replacement therapy (ERT) necessitate early diagnosis because this change the natural history of the disease Few patients have mutations that prevent to synthesize any enzyme and are label CRIM negative. This is a poor prognostic factor. CRIM negative patients required immunomodulation therapy prior starting ERT. \textbf{AIM:} To present an infantile PD, CRIM negative patient that started ERT at 40 days of life. Case 17 months of age male patient product of the second normal pregnancy of healthy non-consanguineous couple. Prenatal diagnosis for PD showed the same mutations as his older sister who have died when she was 1 year of age and she had deleterious mutations in the GAA gene that were known as severe: CRIM negative. Patient prenatal echocardiogram showed hypertrophic cardiomyopathy. The newborn echocardiogram confirm the cardiomyopathy and plasma CPK was very high. The patient receive immune tolerance induction with rituximab, methotrexate, and gamma globulins. ERT started at 40 days of life. He has not presented until now adverse effect. Antibodys have been negatives. The clinical picture of the patient is completely different from that of his sister. He sat down at 7 months, achieved walking at 13 months, he is able to stand up from ground alone, and he has several words and normal behavior. \textbf{CONCLUSIONS:} ERT is beneficial for PD classical infantile. It is very important to think about this diagnosis because as early the confirmation the greater the benefits of this therapy. CRIM negative patients required immunomodulation therapy prior starting ERT.