First successful concomitant therapy of immune tolerance induction therapy and desensitization in a CRIM-negative infantile Pompe patient

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Objectives: Enzyme replacement therapy (ERT) with alglucosidase alfa (rhGAA) has changed the fatal course of infantile Pompe disease, however, development of anti rhGAA antibodies and infusion-associated reactions (IAR) restrict the tolerability and effectiveness of the treatment.

Case presentation: We describe a successful concomitant immune tolerance induction (ITI) and desensitization protocols in a cross-reactive immunologic material (CRIM) negative 7-month-old male patient. At the age of 5 months and eighth dose of the ERT, the patient developed IAR and his rhGAA specific IgE was negative however, his rhGAA specific IgG titer was as high as 12,800. ITI therapy to suppress antibody formation and a desensitization protocol was devised to be given concomitantly. At the end of 5-week therapy, his fatigue and weakness improved profoundly and a control antidrug antibody level decreased at 800. At the time of the patient’s follow up, he was still on ERT with desensitization at the age of 15 months without any reactions.

Conclusions: This is the first report in the literature applying concomitant ITI and desensitization protocols in a CRIM negative infantile-onset Pompe disease patient successfully, hence the importance of the case.