INTRODUCTION

Beta-thalassemia major is a severe form of beta thalassemia with an early onset. It is a common genetic disorder in Mediterranean countries and it is caused by mutation in beta globin gene. Notwithstanding that it is risky, alloegenic hematopoietic stem cell transplantation (HSCT) is currently the only treatment that is effective cure for beta thalassemia. Congenital hypothyroidism is also a condition resulting with deficiency of thyroid hormone in newborn infants. Autoimmune thyroid dysfunction in children with thalassemia major is very rare and poorly described. We report a case of a 13 y/o child with clinical hypothyroidism diagnosed in early childhood, and beta thalassemia major who developed autoimmune thyroiditis with unusual subacute thyroid dysfunction.

CASE PRESENTATION

On presentation 13 years old male with anterior neck pain, fever that lasted for 5-6 hours, cramp-like pain, abdominal bloating and mild diarrhea.
- Past medical history significant for beta thalassemia major and clinical hypothyroidism diagnosed during the infantile period. He had received regular L-thyroxine therapy and also multiple transfusions together with chelation therapy. At the age of 11 years, haploidentical transplantation of hematopoietic stem cells from his mother was performed as well as splenectomy. Since then, patient was put on immunosuppressive therapy.
- The patient was admitted and full clinical examination was performed.
- Blood pressure - 80/60mmHg.
- His weight and height were at the 75th and 50th percentiles, respectively.
- The laboratory evaluation of the thyroid function revealed:
  - increased FT4 levels 5.9 ng/dL (normal reference range 0.9 – 1.4 ng/dL)
  - decreased TSH levels 0.029uU/mL (normal reference range 0.400-4.00 ulU/mL) (explaining the thyrotoxic crisis with hormonal discharge)
  - Extremely elevated the levels of thyroglobulin antibodies (2168 IU/mL: normal, <20)
- The ultrasound revealed normal thyroid size, shape, and contour with hypoechoic structure. The pseudosial was positive for Adenovirus IgG.
- Acute autoimmune thyroiditis due to the viral infection in immunocompromised child was diagnosed.
- Based on these findings, the thyroxine replacement therapy that the child was receiving was temporarily stopped.
- Consequently, the patient was monitored, without any clinical manifestation appeared.
- After several weeks the therapy was given gradually

- TSH level become normal – 0,159uU/mL.
- Free T4 was lower (2,03 ng/dL)
- Anti TPO was 76,9 IU/mL
- TG antibody titer persisted to be elevated > 3000 IU/mL.
- He was discharged with recommended oral prophylactic phenoxymethylpenicillin (penicillin V) therapy of 1000 mg, once daily
- He still attends regular controls for his thyroid status.

CONCLUSION

Although the simultaneous occurrence of beta thalassemia major, acute and autoimmune thyroiditis in patient on thyrroxine replacement therapy can be coincidental, this combination is rare in childhood. To the best of our knowledge, there is no evidence about a plausible association between the acute autoimmune thyroiditis and immunosuppressive therapy given after transplantation. This type of thyroiditis can be overlooked and early diagnosis is important to correct the negative systemic effect of thyroid dysfunction, so clinicians should consider this possibility. Follow up of thyroid function in transplanted children that have received immunosuppressive therapy is mandatory.

REFERENCES