

Follow-up and management of endocrine and metabolic disorders after hematopoietic stem cell transplantation in a patient followed for Fanconi anemia

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1 INTRODUCTION

Fanconi anemia (FA) is a very rare, complex and chronic genetic disorder. The diagnosis is most often made at pediatric age. Hematopoietic stem cell transplantation (HSCT) is the only curative treatment for bone marrow failure. FA on its own as well as its treatment can affect the endocrine system. We report the case of a child followed for 12 years after HSCT for FA.

2 OBSERVATION

A patient has been followed since the age of 7 for FA complicated by aplastic anemia. She had an allogeneic bone marrow transplant when she was 8 years old. During her follow-up, the diagnosis of GH deficiency was made at the age of 10 years and the patient had treatment with growth hormone for 2 years. She had had chronic kidney failure since the age of 13 with peritoneal hemodialysis and calcium and vitamin D supplementation. She had menarche at the age of 13 with regular periods.

Currently she is 20 years old. On physical examination, her final height was 1.36 m with a BMI of 24.4 kg / m². She had stage 3 heterogeneous goiter with no signs of hypothyroidism. In biology, the synacthen test was normal. TSH was 16.4 μ IU / L and FT4 was 6 ng / L (VN: 9-19) controlled at 0.05 μ IU / L and 9.3 ng / L respectively. Anti-peroxidase antibodies are negative. She had prediabetes with hypertriglyceridemia and hypercholesterolemia. The pituitary MRI is without abnormality. She was put on L-thyroxine and dietetic rules.

3 DISCUSSION

The GH deficiency in our patient would most likely be secondary to total body irradiation (TBI) during the HSCT preparation protocol. The final height was very delayed which could be further explained by the FA. Some mutations are more at risk of small stature, independent of hormone levels.

FA is not a contraindication to treatment with GH, but treatment with GH should be temporarily interrupted before the HSCT and for at least 6 months after the transplantation, as well as in serious illnesses.

Interpretation of the thyroid workup was difficult in our patient because hypothyroidism could be either secondary to autoimmune thyroiditis post HSCT, a diagnosis invalidated by the negativity of ATPO, or secondary to thyrotropic insufficiency due to TBI.

Interpreting the metabolic balance is tricky. Dyslipidemia could be of multifactorial origin: Chronic renal failure, FA or secondary to HSCT. Indeed, even after the interruption of immunosuppression, allogeneic HSC transplant patients seem to have an increased risk of diabetes and metabolic syndrome.

4 CONCLUSION

At present, allogeneic HSCT remains the only treatment that corrects the haematological complications common to most patients with FA. All patients who have received radiochemotherapy and an allogeneic HSCT are likely to develop late complications. Endocrinological and metabolic evaluation is necessary. A multidisciplinary team caring for these patients should include a pediatric or adult endocrinologist and work closely with other FA specialists to ensure appropriate and optimal patient care.

