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Case Report

Challenge Treatment GH with Turner Syndrome Having Cardiac and Renal Malformation: About A Case Report

Hassan Aden Neima[1*], H. Ouakrim[1], S. Rafi[1], G. El Mghari[1], N. El Ansari[1]

[1]Department of Endocrinology, Diabetes, Metabolic Diseases and Nutrition, Mohammed VI University Hospital, Marrakech, Morocco

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***Corresponding Author:** Hassan Aden Neima
Department of Endocrinology, Diabetes, Metabolic Diseases and Nutrition, Mohammed VI University Hospital, Marrakech, Morocco

Abstract

In GH deficiency, rhGH is deemed appropriate when epiphyses are open, while Turner's syndrome poses unique considerations due to associated malformations like cardiac aortic bicuspidity and aortic coarctation. Renal malformations, including Horseshoe kidney and hydronephrosis, further highlight the complexity of Turner's syndrome, requiring a comprehensive, multidisciplinary approach to care. We report the case about of an elderly child 3 years and 5 months old patient with cardiac and renal malformations notably a tight postductal isthmic coarctation and a left pyeloureteral junction syndrome with dilatation of the pyelocalic cavities, admitted for further management of a staturo-ponderal delay. She presented with functional signs of constipation, no chills, no digestive disorders and no hypoglycemic malaise. On physical examination, dysmorphic features included low hairline and short neck, pterygium coli, low ear implant, hypertelorism, nipple spread, keloid lesion on face. Test results showed a 45x karyotype in favour of Turner syndrome, HBA1c: 5.6%, the rest of the tests were normal. GH was indicated in the patient after cardiac surgery for coarctation of the aorta and pyelo-caliceal junction. The patient was referred for heart and kidney surgery. It is crucial to closely monitor height velocity during catch-up growth, particularly in patients with concurrent conditions leading to short stature. Additionally, there is an emphasis on educating parents about the potential impact of Turner syndrome on aspects such as growth, growth hormone (GH) substitution, ovarian failure, malformations, and related health risks.

Keywords: GH, malformation cardiac and kidney, turner syndrome.

INTRODUCTION

Recombinant human growth hormone (rhGH) serves as the primary treatment for short stature induced by growth hormone (GH) deficiency, addressing associated abnormalities in body composition, metabolic profile, exercise capacity, and quality of life. Challenges in effective treatment include the difficulty in establishing a definitive diagnosis of GH deficiency and variable responsiveness to rhGH within the diagnosed population.

This review focuses on the indications for and efficacy of exogenous rhGH treatment in children with GH deficiency, detailing the diagnostic approach to children with short stature and the confirmation of GH deficiency. Additionally, rhGH therapy is prescribed for various specific indications, including Turner syndrome, a rare chromosomal disorder linked to the total or partial absence of an X chromosome. Diagnosis is confirmed through a karyotype, with monosomy 45, X being one form of Turner syndrome. This syndrome typically involves statural retardation and ovarian failure, along with a variable dysmorphic syndrome and an increased risk of congenital malformations, particularly cardiac and renal anomalies.

For pediatricians, general practitioners, and school doctors, considering Turner syndrome in cases of statural deficit (height ≤ - 2DS), even if growth remains regular, is crucial for facilitating an earlier diagnosis and appropriate management of Turner syndrome

CASE PRESENTATION

A 3-year and 5-month-old patient with cardiac and renal malformations, notably a tight postductal isthmic coarctation and a left pyeloureteral junction syndrome with dilatation of the pyelocalic cavities, was admitted for further management of staturo-ponderal delay. She exhibited functional signs of constipation, with no chills, digestive disorders, or hypoglycemic

malaise. On physical examination, dysmorphic features included a short neck, low implanted hair, pterygium coli, low ear implant, hypertelorism, nipple spread, and a keloid lesion on the face. Test results revealed a 45x karyotype indicative of Turner syndrome, and thoracic angioscanner displayed a tight aortic coarctation. HbA1c was measured at 5.6%, while the rest of the tests yielded normal results. Growth hormone (GH) was indicated in the patient after cardiac surgery for coarctation of the aorta and pyelocaliceal junction. Subsequently, the patient was referred for heart and kidney surgery.

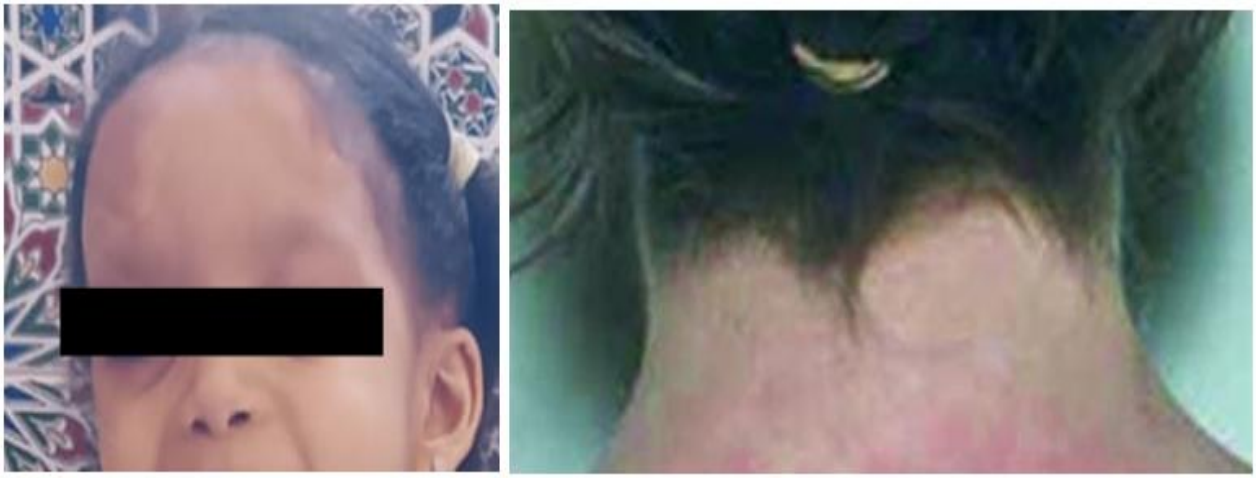

Figure 1: Syndrom dysmorphic of turner

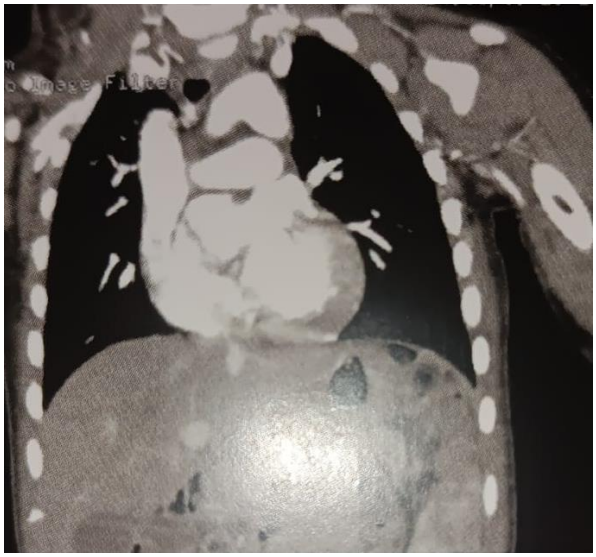

Figure 2: Thoracic angioscan for aortic coarctation

DISCUSSION

Treatment with recombinant human growth hormone (rhGH) is appropriate for children with GH deficiency whose epiphyses are open [1]. Turner's syndrome is also an indication for GH treatment, considering malformations such as cardiac aortic bicuspidity and aortic coarctation [2]. In the kidney, malformations like Horseshoe kidney, duplication of the collecting system, and hydronephrosis may occur. These malformations are often symptomatic and should be investigated by renal and cardiac ultrasound when diagnosing Turner syndrome, necessitating a multidisciplinary approach to care. Such malformations are risk factors for aortic dilatation and may predispose to aortic dissection [3].

The phenotype of Turner syndrome patients is influenced by X chromosome gene dosage and other phenomena, such as altered regulation of gene expression triggered by the absence of the second sex chromosome [4]. An American registry of Turner syndrome patients with a history of GH treatment (5520 patients followed for 20 years) found no significant association between GH treatment and aortic dilatation or aortic dissection [5]. Following cardiovascular surgery in patients with Turner syndrome and on GH, the long-term survival was 98%.

Factors associated with a better response to GH treatment include greater height at treatment initiation, greater parental height, younger age at treatment

initiation, and duration of treatment. The recommended dose of GH varies between 0.045 and 0.050 mg/Kg/d1. Growth hormone treatment is discontinued when bone age is greater than or equal to 14 years, and growth rate is less than 2 cm per year [7]. Therapeutic education is integral to the management of patients with Turner syndrome.

Observational studies on large cohorts show a greater propensity of these patients to develop transient intracranial hypertension at the start of GH treatment compared to patients without Turner syndrome. The growth response is greater when rhGH is initiated at a younger age. Therefore, treatment should be initiated as soon as the diagnosis is confirmed and continued until linear growth ceases.

CONCLUSION

In patients with a comorbid disease contributing to short stature, such as Turner syndrome, after prescribing rhGH, height velocity is monitored at three- to six-month intervals. The goal for treatment is to attain the 75th percentile for height velocity for age during catch-up growth. Parents must be well-informed about the consequences of Turner syndrome on adult growth and height, GH substitution, ovarian failure, the potential risk of malformations, and associated comorbidities, as well as screening and management recommendations.

Compliance with ethical standards

Acknowledgments

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Disclosure of conflict of interest: The authors declare no conflict of interests.

Statement of ethical approval

The present research work does not contain any studies performed on animals/humans subject by any of the authors.

Statement of informed consent: Informed consent was obtained from all individual participants included in the study.

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