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Intensive Treatment with Alfa-Iduronidase decreases Complications in "Hurler-Scheie Syndrome" Patients requiring Surgery: A Case Report

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Abstract

Mucopolysaccharidosis type I (MPS I) is a rare lysosomal storage disease that leads to accumulation of glycosaminoglycans (GAGs) in different tissues. Respiratory insufficiency is the most important cause of death in patients with this disease, especially during pre/post anesthesia. We present the first report of the use of alfa-iduronidase in an intensive regimen to prevent respiratory complications during surgery in a case of Hurler-Sheie syndrome. An intensive treatment with alfa-iduronidase is proposed and shown to be effective in the prevention of respiratory complications in patients with MPS I that require surgery.

Keywords: Hurler-Scheie; Surgery; Alfa-iduronidase

Introduction

Mucopolysaccharidosis type I (MPS I) is a rare lysosomal storage disease that leads to accumulation of glycosaminoglycans (GAGs) in different tissues and organs [1,2]. This accumulation leads to a progressively worsening of the organs functions and a decrease the lifespan. MPS I can be subdivided in to three phenotypes of increasing severity: Scheie, Hurler-Scheie and Hurler. Patients with MPS I are treated with weekly infusions of alfa-iduronidase (Aldurazyme®), the enzymatic substitute for this disease, in an attempt to reduce GAG accumulation and ameliorate chronic symptoms [3]. Respiratory complications are common due to soft tissue involvement in the respiratory pathways. Airway obstruction after induction or extubation difficulties are frequent, and many patients require tracheostomies which are not always effective [4]. Respiratory insufficiency and its complications is the most important cause of death in patients with this disease, especially during pre/post anaesthesia [1,2,5]. We present the first report of the use of alfa-iduronidase in an intensive regimen to prevent respiratory complications during surgery in a case of Hurler-Sheie syndrome.

Case Report

We present a patient of 27 years suffers from "Hurler-Scheie syndrome" treated with recombinant alfa-iduronidase at a dose of 0.58 mg/Kg in weekly infusions. At the age of 22 years he required surgical intervention because of severe scholiosis. Surgery was uneventful but he could not be extubated and required a tracheostomy. When consulted by the intensive care specialists, and in an attempt to reduce soft tissue infiltration and ameliorate his prognosis, we decided to give him his alfa-iduronidase infusión early, only 5 days after the previous dose. Two days later he was able to be extubated [4]. He then resumed his usual weekly schedule of infusions, had no further complications and could be discharged from hospital. He lead his usual life for 4 years but then required a second intervention for his scholiosis (Figure 1). In this second occasion we intended to prevent respiratory complications and decided, with the informed consent to the patient, to give him an infusion at his regular dose 72 hours prior to surgery and a second dose on the day of the intervention. He suffered no complications, which allowed an early extubation 24 hours after surgery and discharge from the hospital on day 5 with his regular treatment. No side effects were observed due to the higher frequency of infusions in neither occasion.

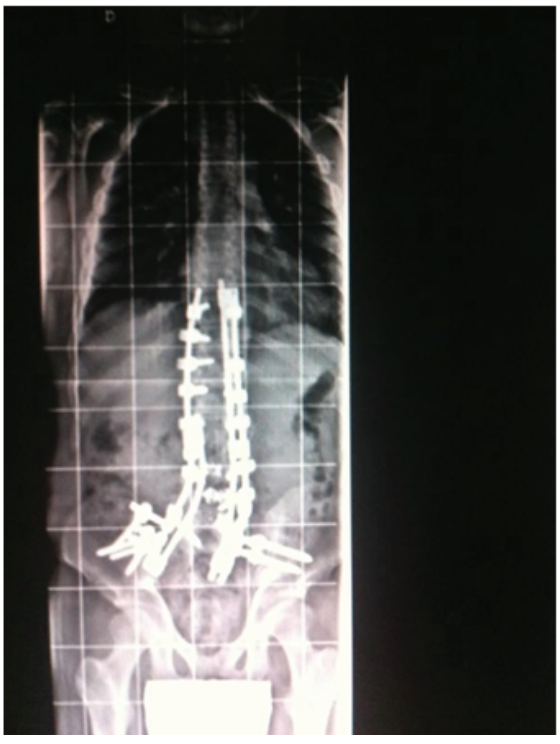

Figure 1: Intervention for scholiosis.

Discussion and Conclusion

Patients with MPS have high prevalence of airway obstruction and restrictive pulmonary disease, which lead to their premature death. Any intervention on these patients can worsen their basal condition and it is of high risk due to respiratory complications [6]. GAG accumulation in the upper airway can even impede visualization of the glottis and therefore it is not surprising that the most frequent problems of anaesthesia include airway obstruction after induction, extubation/intubation difficulties or failure of intubate [7].

However, patients who have Hurler-Scheie síndrome have bone abnormalities that could often benefit from surgery which is not undertaken due to the risk of complications. In situations of spinalcord compression at cervico cranial and thoraco lumbar regions such as our case, surgery is indispensable but it is performed only as a last measure.

Substitute enzymatic therapy is usually recommended to avoid chronic complications of the different mucopolysacharide deposit diseases, and treatment is implemented at a regular dose in standard intervals. In this patient we previously proved that an emergency treatment could be of benefit once complications have arisen [4]. This is the first report in which an intensive treatment with alfa-iduronidase was proposed and demonstrated to be effective in the prevention of respiratory complications in patients with MPS I that require surgery.

Apart from our experience, it has not been previously suggested in humans that the substitutes dose could be of benefit in emergency situations such as surgery. Preanesthetic evaluation of these patients could help identify those at themostrisk of complications in this and other mucopolysacharide diseases [6-10], in order not only to prepare for posible complications but also determine those who might benefit from an intensive substitutes regimen, as it was proven by our case.

Conflict of Interest

The authors declare that they have no competing interests.

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