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Anesthesia management of an infant with artemis severe combined immune deficiency

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Abstract

Artemis is a nuclear protein that plays an essential role in DNA repair and was first identified as the gene defective in a subset of severe combined immune deficiency patients that were unusually sensitive to radiation. Management of patients with severe combined immune deficiency may be challenging. The disadvantage of Chest X Rays and CT scan is exposure to radiation which was an inacceptable approach for this patient. The disadvantages of MRI include the length of the exam and the need for sedation in pediatric patients in order to achieve immobility. Risks of anaesthesia are increased in the presence of upper airway and/or pulmonary disease, and more so in patients with SCID secondary to risk of iatrogenic infection risk and severe complications. The feed and sleep technique may succeed in avoiding the risks of general anaesthesia and sedation in these high-risk infants.

Introduction

Artemis is a nuclear protein that plays an essential role in DNA repair and in V (variable), D (diversity) and J (joining) recombination: a process by which B cell antibody genes and T cell receptor genes are assembled. In Artemis-deficient individuals, no mature B or T cells are produced, a condition known as severe combined immune deficiency (SCID). Artemis was first identified as the gene defective in a subset of SCID patients that were unusually sensitive to radiation [1-4].

Case presentation

Sedation was requested for a full term 5-month-old infant, 6.3 kg, as a late afternoon add on case for neck and chest magnetic resonance imaging (MRI). Patient had symptoms of respiratory infection including rhinorrhea and cough, in addition to torticollis. Patient's newborn screen came back critical positive for T negative, B negative, natural killer cell positive severe combined immunodeficiency, with sensitivity to ionizing radiation. Genetic testing confirmed the diagnosis of a mutation in DCLRE1C – which codes the Artemis protein. An unrelated match was found, and the patient was scheduled for bone marrow transplant as definitive cure for SCID. After discussion with the MRI ordering physician, there was a request to anesthetize the patient as soon as possible to avoid any delay in the management and care of this patient. After evaluation by the anesthesiologist, the infant was fed just prior to the MRI and allowed to fall asleep naturally with ears covered with earmuffs and bundled in an MRI-immobilizing swaddler and there was a discussion with the radiologist to carefully review the length of the scan to ensure gathering of the best possible information in the shortest amount of time. The procedure was safely performed with the capacity of gathering the information needed from the MRI scan.

Discussion

Management of patients with severe combined immune deficiency may be challenging. The disadvantage of Chest X Rays and CT scan is exposure to radiation which was an inacceptable approach for this patient. The disadvantages of MRI include the length of the exam and the need for sedation in pediatric patients in order to achieve immobility. Risks of anesthesia are increased in the presence of upper airway and/or pulmonary disease, and more so in patients with SCID secondary to risk of iatrogenic infection risk and severe complications. The feed and sleep technique may succeed in avoiding the risks of general anesthesia and sedation in infants, and is worth trying, especially in high risk patients, even if it does not coincide with the infant's sleep or feeding patterns. The disadvantage of the feed and sleep technique in case of failure, is waiting to meet appropriate NPO status. Histories of preterm birth and spine MRI have been reported to be associated with a less-successful scan outcome in infants of three months of age or younger. 79% of scan using the feed and sleep technique addressed the clinical question, 20% partially addressed the clinical question [5]. Communication with the different teams involved in caring for patients with severe combined immune deficiency is the best approach for a safe outcome (Table 1).

Table 1. Lymphocyte Phenotype of selected SCID types

Gene Mutation	NK cell	B cell	T cell
ADA deficiency	-	-	-
Common gamma chain deficiency	-	+	-
CD3$^{\delta}$	+	+	-
CD45	+	+	-
Artemis deficiency	+	-	-
Omenn Syndrome	+	-	-
Ligase 4 deficiency	+	-	-
JAK3	-	+	-
IL-7RGα	+	+	-

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Conclusion

In patients with diagnosis of Artemis Severe Combined Immunodeficiency, careful management should be focused on avoiding radiation and infection by providing the safest and least risk associated sedation/ anaesthetic approach.

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