

## INTRODUCTION

"RYR1-related disorders" is a broad term encompassing various mutations within the ryanodine receptor 1 (RYR1) gene that impacts the neuromuscular system in humans. These mutations are the primary cause of congenital muscle diseases. The RYR1 receptor, found in muscle cells, plays a crucial role in regulating calcium flow, essential for muscle contraction. Dysfunctional muscle contractions result from either a reduced number or abnormal function of RYR1 channels. Clinically, individuals with RYR1-related disorders exhibit symptoms such as muscle weakness, atrophy, hypotonia, hyporeflexia, and delayed motor development. Breathing difficulties associated with these disorders range from mild to severe, stemming from weakened chest wall muscles. Mild cases may manifest as sleep apnea necessitating breathing support during sleep, while severe cases require continuous assistance. Additionally, RYR1 variants are the primary cause of malignant hyperthermia, accounting for over 60% of cases. Malignant hyperthermia is a potentially fatal reaction triggered by exposure to certain volatile anesthetics or depolarizing muscle relaxants, leading to rapid hyperthermia and muscle breakdown in susceptible individuals.

## CASE REPORT

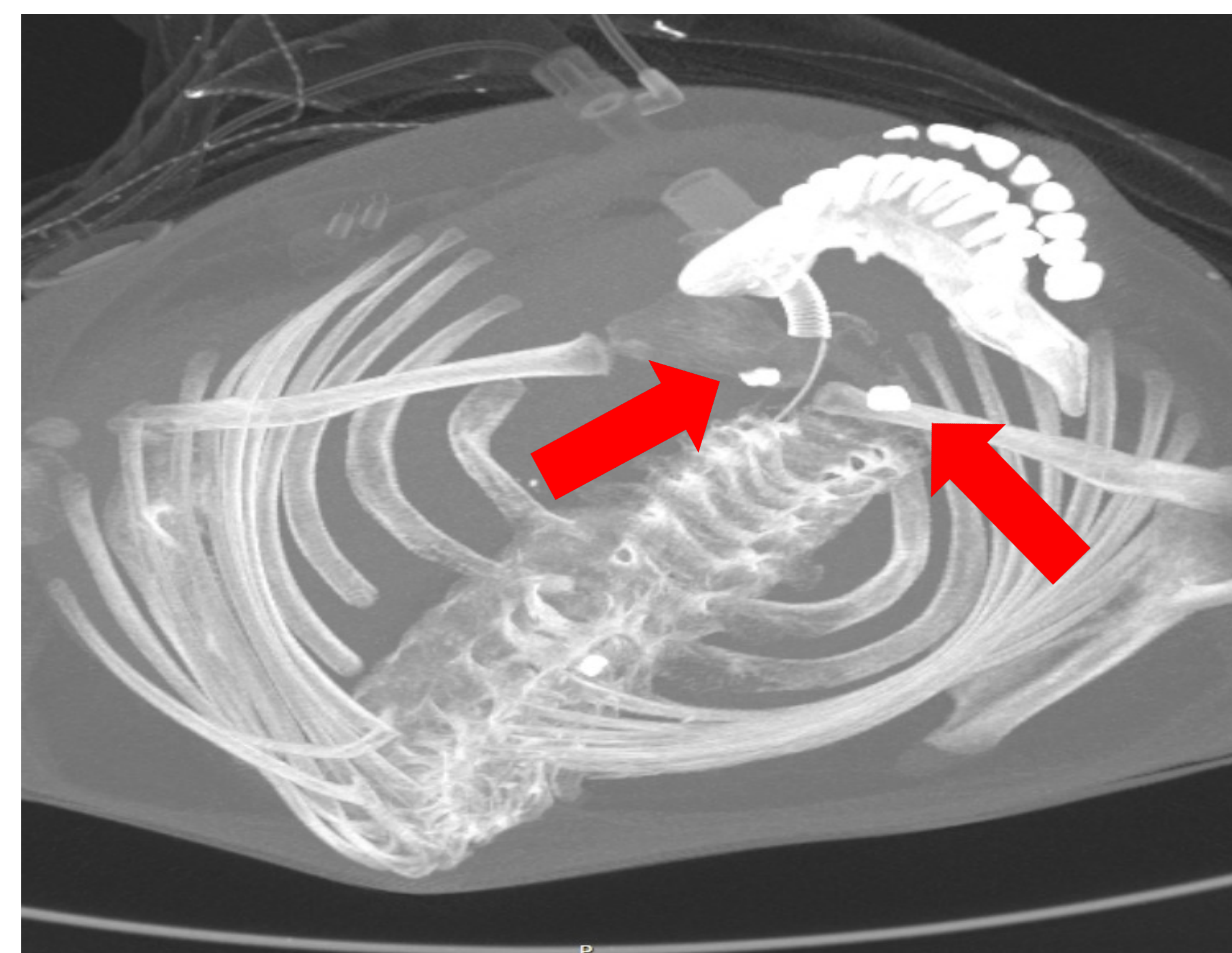
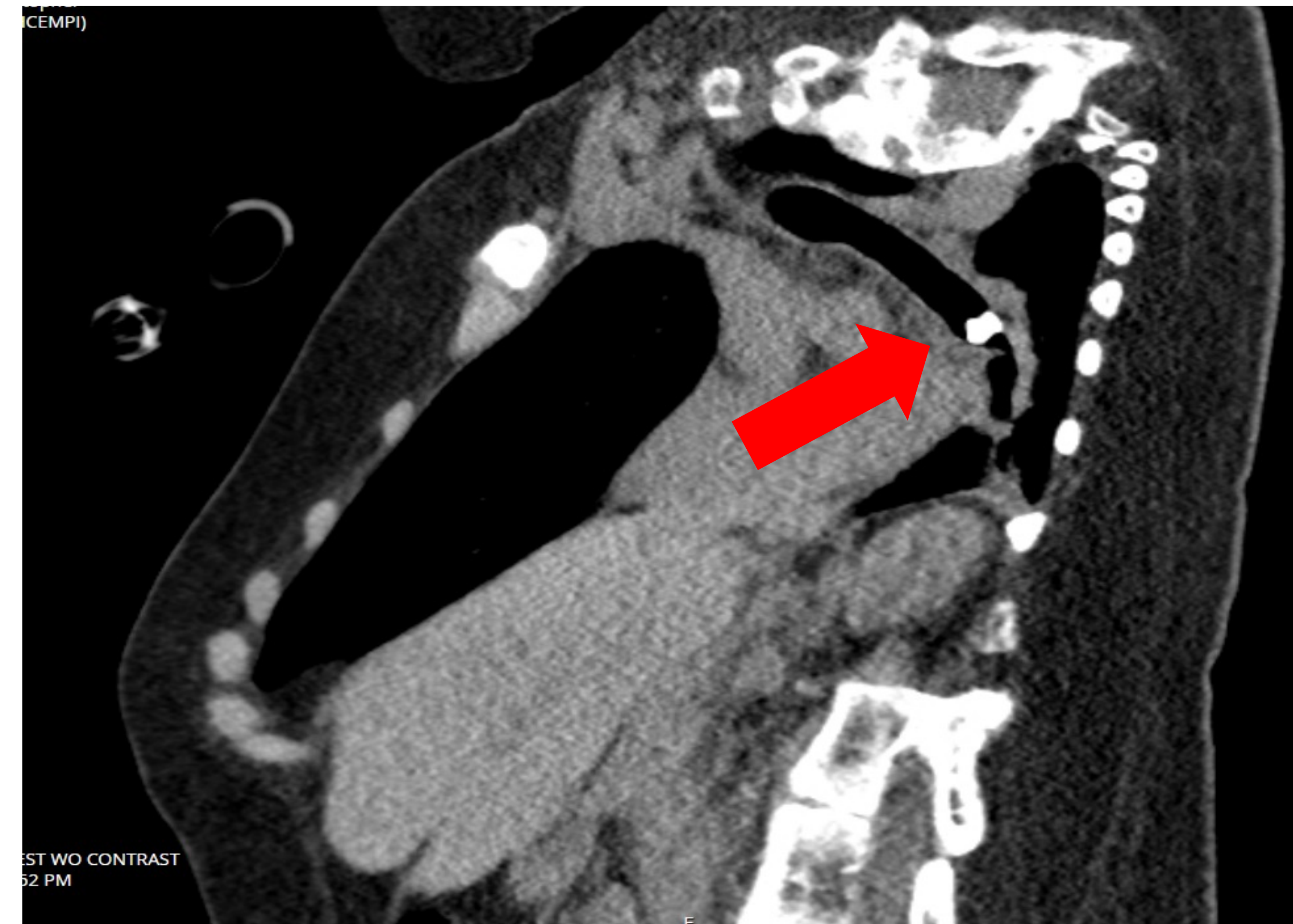
A 10-year-old male patient was referred to University Hospitals Rainbow Babies and Children's Hospital after a routine chest exam detected one primary tooth in the left main bronchus and two primary teeth in the pyriform sinus.

This 10-year-old patient's medical history is significant for chronic respiratory failure due to RYR1 mutation with subsequent tracheostomy and ventilator dependence, congenital myopathy, congenital scoliosis, G-tube dependence, neurogenic dysphagia, and arthrogryposis. His current medications are albuterol with IPV (intrapulmonary percussive ventilation), flovent inhaler, and azithromycin. No reported drug allergies. This patient has a dental home at an outside office but has not been seen by a dentist in over two years.

The patient was seen by pediatric otorhinolaryngology, pediatric pulmonology, and pediatric dentistry under general anesthesia (GA) for the retrieval of the aspirated teeth and a dental exam. While under GA, the patient had severe trismus restricting access to the oral cavity with ENT instrumentation. Pediatric pulmonology then completed bronchoscopic removal of the tooth from the left main bronchus and used interventional techniques to remove the two teeth in the pyriform sinus. The dental team then conducted a comprehensive oral exam. The clinical and radiographic exams revealed full permanent dentition with over-retained primary tooth A. A dental prophylaxis was completed and the remaining primary tooth was extracted to avoid future aspiration.

## RADIOGRAPHIC PRESENTATION

- CT Chest Scan April 2023



## DISCUSSION

This case report discusses a 10-year-old male with an RYR1 mutation, in whom three aspirated primary teeth were incidentally discovered during a routine chest examination. Due to his severely weakened chest wall muscle function, he is ventilator dependent and non-verbal – leaving this patient unable to communicate discomfort. Treatment required a specialty team of pediatric otorhinolaryngology, pediatric pulmonology, and pediatric dentistry to retrieve and extract the remaining primary teeth under GA. This case highlights the importance of parents and doctors remaining vigilant during times of primary teeth exfoliation in children with RYR1-related diseases. These patients should undergo routine dental exams as a precautionary measure. If primary teeth exhibiting mobility secondary to exfoliation are discovered during an exam, extraction of these teeth are recommended for patients with RYR1-related diseases, in addition to other neuromuscular disorders, such as cerebral palsy, multiple sclerosis (MS), amyotrophic lateral sclerosis (ALS), and Guillain-Barré syndrome. In cases where primary teeth are unable to be found post-exfoliation, a chest x-ray would be recommended. It has been noted that “[c]onsequences of foreign body aspiration include acute respiratory compromise, pneumonia, and can lead to death” [5]. With such severe potential complications, dentists have an important role in the prevention of aspirated primary teeth.

## REFERENCES

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