[NM-270] Unanticipated difficult intubation in a pediatric patient with an Xp22.3 Duplication

Lee K, Tassone R, Changyaleket B
University of Illinois Hospital and Heath Sciences , Chicago , IL, USA

Duplications of the short arm of the X chromosome in male patients are rare. A comprehensive description of the Xp22.3 duplication phenotype is still not well defined. Previous descriptions of patients with similar duplications show that they typically have intellectual disabilities, facial dysmorphic features, seizures, and other variable clinical problems. We present the first known case of a difficult intubation in a child with an Xp22.3 duplication.

Our patient is a three-year old boy who presented for complete oral rehabilitation. His past medical history includes an Xp22.3 duplication, brief seizures during infancy, developmental delay, hypothyroidism, and subtle facial dysmorphic features. The patient's parents report that he started walking late at the age of three. He communicates with gestures and has not spoken any words. The patient's facial features include frontal bossing, epicanthal folds, depressed nasal bridge, and macrocephaly.

During nasal intubation, initial direct laryngoscopy with a MAC 2 allowed view of only epiglottis because the blade was too short. A WIS 1.5 was then used with view only of the epiglottis. The patient began to desaturate and he was mask ventilated until oxygen saturation returned to 100%. While waiting for the video laryngoscope, a third DL attempt with a WIS 1.5 provided a view of arytenoids. The endotracheal tube was aimed superior to the arytenoids and was advanced into the trachea with some difficulty. Although the patient had mild facial dysmorphic features a difficult intubation was not readily expected.

As genetic testing advancement continues more patients are being diagnosed with previously unknown genetic abnormalities. X-linked intellectual disability includes both syndromic and non-syndromic patients. Li et al. reported that out of 7793 microarray samples from patients with suspected syndromic features, 29 individuals had microduplications found at Xp22.31 [1]. As Xp22.3 microduplications are rare genetic abnormalities the majority of the current literature are case studies. The available studies show a propensity for intellectual disability and facial dysmorphic features [2-3]. Our case study is the first reported difficult intubation of a pediatric patient with an Xp22.3 duplication. As more of these rare duplications in the short arm of the X chromosome patients are described in the literature, a greater understanding of appropriate anesthesia care can be defined.

1. Li F et al. 2010. Interstitial microduplication of Xp22.31: Causative of intellectual disability or benign copy number variant? Eur J of Med Gen. 53(2):93-9
2. Tejada M et al. 2011. A child with mild x-linked intellectual disability and a microduplication at xp22.12 including RPS6KA3. Pediatrics. 128:1029-1033
3. Liu P et al. 2011. Copy number gain at Xp22.31 includes complex duplication rearrangements and recurrent triplications. Hum. Mol. Gen. 20:1975–1988