# The Anesthetic Management and Implications of Kniest Syndrome

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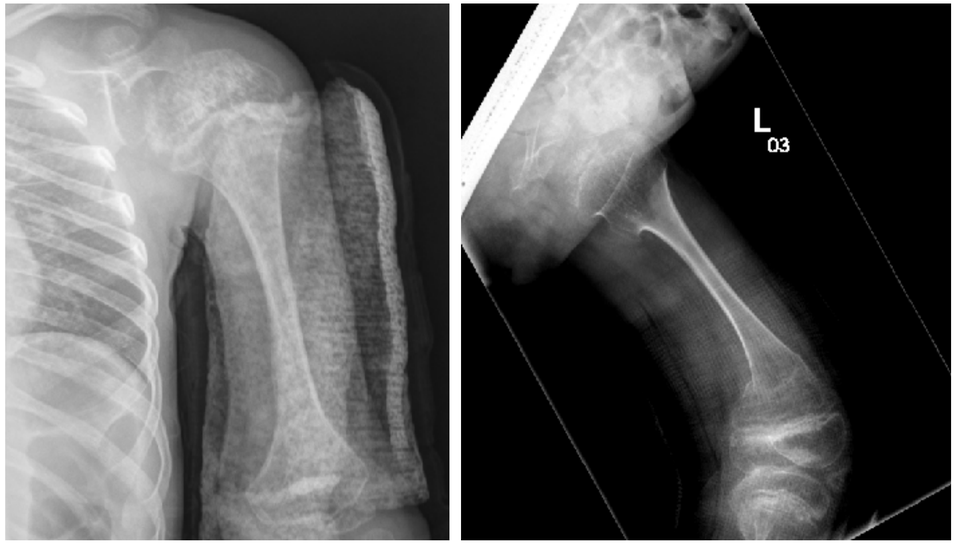
## INTRODUCTION

Kniest dysplasia is an "atypical chondrodysplasia," caused by mutation of the COL2A1 gene which encodes type II collagen. Patients exhibit craniofacial abnormalities such as macrocephaly, flat midface and nose root, and cleft palate. Abnormal tracheal cartilage may result in tracheomalacia. Ventilation and perfusion usually remain unaffected, despite a short, narrow chest. We present a patient with a history of Kniest

## CASE REPORT

A 6 year old male presents with mitochondrial disorder, skeletal deformities, infantile spasms, severe developmental delay, asthma and bilateral cataracts for an eye EUA prior to cataract surgery. His mother described a "catastrophic allergic reaction" to previous anesthetic including propofol and sevoflurane, causing irreparable neurologic damage. Physical exam revealed a 12.5 kg nonverbal wheelchair-bound male with contractures and small mouth opening. After placement of ASA monitors and pre-oxygenation, an IV was placed in the OR with an ultrasound. 12 ug dexmedetomidine and 25 mg of ketamine were given to induce. A 3.5 uncuffed ET tube was placed following multiple failed attempts with an appropriately sized ET tube; subglottic stenosis was suspected. The patient was paralyzed with rocuronium and maintained on dexmedetomidine infusion. After uneventful extubation he was discharged home that day, subsequently undergoing cataract surgery successfully with the same anesthetic plan.




Long bone x-rays showing the characteristic dumbbell shape

## DISCUSSION

Kneist syndrome patients present multiple anesthetic challenges. They are at risk for atlantoaxial instability. Head and neck stabilization during intubation and refraining from severe respiratory depression is recommended. Respiratory failure is contingent on the degree of kyphoscoliosis. Growth retardation is common. Large deformed epiphyses, broad metaphyses and short tubular bones result in a "dumbbell" shape, seen in attached figures. Joint space is narrowed with limited mobility. Early-onset myopia, retinal detachment, and sensorineural hearing loss are common. Fixed flexion of major joints make IV access and positioning a challenge. Considering the multiple connective tissue abnormalities related to airway management, difficult intubation can be anticipated. Our patient is a unique case of Kniest dysplasia with multiple anesthetic allergies. Knowledge of comorbidities and a good H&P are paramount in delivering a safe anesthetic.

## REFERENCES

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