

Medical Alert for Parents of Children with I-Cell Disease and the Physicians Who Care for Them

I-Cell disease is a slowly progressive inborn error of metabolism for which, unfortunately, no curative treatment is available. Children with I-Cell disease have extreme growth failure, neurodevelopmental delay and disease of connective tissues in many organs, including the lungs and respiratory tract. Manifestations including swollen gums, stiff joints, thick-feeling skin, bone and tendon abnormalities are well known signs of the disorder.

Because of the poor growth potential, atrophied muscles, and poor compliance of the narrow thoracic cage, the baseline respiratory status of these children is compromised. Additionally, they have small mouths that do not open fully or easily. As the children grow older the airway becomes gradually more restricted as the initially swollen airway passages become stiff and easily traumatized. This combination of factors makes intubation (placing a tube within the airway in order to use mechanical assistance to breathe for the patient) very risky, so *elective surgical procedures should be avoided as much as possible*.

If a procedure is considered essential, it should be undertaken at a major medical facility where pediatric anesthesia and pediatric critical care services are available. Under all circumstances, medical staff should be prepared to perform fiberoptic intubation, where a tiny camera is used to visualize the airway during the intubation procedure. Because children with I-Cell disease have much smaller airways than other children of the same age, a much smaller endotracheal tube will be required. However, a smaller tube may be less effective for ventilation (breathing for the patient) and may potentially lead to additional problems such as pneumothorax or collapsed lung.

All medical personnel involved with the elective procedure (surgeons, anesthesiologists, and critical care specialists) should be conversant with the clinical features and natural course of I-Cell disease. Parents and physicians must realize that intubation can easily cause more problems than an elective procedure may alleviate.

For many of the same reasons already described, extubation (removing the breathing tube) may also be unusually difficult and the patient's general condition in the first days following it, quite unpredictable. If prolonged intubation is required, a

tracheotomy (making an artificial hole in the trachea through an incision in the neck) will be required. The probability of subsequent adequate healing in order to close the tracheostomy and having the patient breathe without mechanical assistance is slim indeed.

All humans have an airway protective reflex so as to continue to breathe when sleeping or even when artificially sedated. Deep anesthesia diminishes this reflex. Children with I-Cell disease do not necessarily respond differently to anesthesia. However, because of their precarious airway status from the start, the patients may lose that protective reflex very quickly, even from “light sedation”. That is why a medical team must always be prepared for intubation, preferably fiberoptically, if indeed a surgical procedure is considered of crucial importance.

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