[GA1-51] Ethical considerations and Anesthetic management of an 8 yr old with DK Phocomelia who presents for spinal fusion.

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Phocomelia is a rare congenital defect characterized by severe hypoplasia of the long tubular bones commonly attributed to intrauterine Thalidomide exposure. DK Phocomelia, a condition described by von Voss, and later by Cherstvoy, is an AR syndrome characterized by limb abnormalities, thrombocytopenia, urogenital anomalies and encephalocele. Recent data by Bermejo-Sanchez suggests prevalence of 0.62 per 100,000 births. We present a 12 kg, 8 y.o.male with DK Phocomelia, history of encephalocele repair, disorganization of cerebral hemispheres, rudimentary left-sided limbs, and severe scoliosis for redo of a T1-L5 PSF . His mother was persistent on this procedure after learning that a small screw had become loose even though child was asymptomatic. In the previous surgery arterial cannulation was unsuccessful and tibial artery cutdown was performed. More difficulties were feared this time since his limbs had become more engorged with redundant tissue. The surgeon and anesthesia team were hesitant to perform the procedure, as it would add nothing to his current quality of life, but the mother insisted. The day before surgery he underwent IR guided central line placement. Prior to induction of GA, non-invasive BP cuff was used on rudimentary left limb. After IV induction and intubation, as anticipated, we were unable to measure NIBP. Multiple unsuccessful attempts were made at arterial
cannulation but catheter advancement was impossible; during this time a palpable pulse was our only measure of BP. Options were discussed with family and a femoral cut-down was planned. It took surgeons 150 mins to secure tunneled arterial access; which worked intermittently throughout procedure. After surgery, the patient was extubated without issue. With continued medical advancements extraordinarily rare cases are becoming more prevalent. Due to the complexity in anesthetic management for these patients we pose an important question: “Just because we can, should we do these procedures?” Though little is known about life expectancy for our patient, as the oldest known pt was an 8 y.o. male reported by Urioste , it is certain that he will forever remain bedbound. Retrospectively, we have discussed this case and asked ourselves, “What if we were unable to get any blood pressure readings?” How should we have proceeded? In a review of the literature, we have found a few similar cases in which arterial cannulation was unsuccessful. Mukhtar describes an adult phocomelia patient where he was forced to place a TEE probe to monitor hemodynamics. Gurnaney et al reports using penile blood pressure measurement until arterial cannulation was successful. Though these methods seem extreme these cases, both emergencies, required it. But what about the elective, should we also go to the extreme? Does quality of life play a role in how and when we provide anesthesia? Our case highlights the much needed discussion of when and how do we draw the line.

Cherstvoy E, et al. Syndrome of Multiple Congenital Malformations Including Phocomelia , Thrombocytopenia, Encephalocele, and Urogenital Abnormalities. Lancet 1980; 2: 485
Mukhtar K et al. Anaesthesia and orphan disease: Phocomelia –a lesson from the past. European Journal of Anaesthesiology 2012; 29(7): 353-354
