Anesthetic Implications of Pallister-Hall Syndrome

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Introduction: Pallister-Hall Syndrome (PHS) is a rare genetic disorder diagnosed by the presence of hypothalamic hamartoma, imperforate anus, and polydactyly, but other anesthetically relevant manifestations may include mid-facial hypoplasia, bifid epiglottis, and larygotracheal cleft. PHS demonstrates autosomal dominant inheritance, but often presents as a new mutation. It is caused by a defect in transcription factor gene GLI3. We present the case of an eight year old girl known to have this condition whose bifid epiglottis has been asymptomatic.

Case Report: Our patient was an 8 year old female with an interesting history. She was adopted from Cambodia at age 2 ½ years old after having corrective surgery with placement of a colostomy as a neonate for imperforate anus. She had no known history of seizures, visual disturbances or pituitary abnormalities from the hypothalamic hamartoma. She had no history of feeding problems or aspiration from her bifid epiglottis, and no history of problems with prior anesthetics. She presented to our hospital for elective correction of her right foot postaxial polydactyly. The patient was brought to the operating room, where general anesthesia was induced by mask induction. Though the patient did have mild midface hypoplasia, a characteristic of PHS, she was mask ventilated easily while her oropharynx and hypopharynx were fiberoptically examined and photographed detailing her asymptomatic bifurcated epiglottis. Direct laryngoscopy was performed for examination purposes only and a grade one view of the vocal cords was obtained. A laryngeal mask airway (LMA) size #2.5 was inserted and fiberoptic examination of the epiglottis was also performed through the LMA. The patient then had her orthopedic procedure performed successfully and without complication.

Discussion: Judith Hall, a Canadian geneticist, and Phillip Hallister, and American physician, initially identified the syndrome in 1980, which was based on the presence of a hypothalamic hamartoma, imperforate anus, and postaxial polydactyly (1). Other anomalies now classically associated with PHS include abnormal lung lobulation, midface hypoplasia, laryngotracheal cleft, dysplastic nail beds, renal abnormalities, bifid epiglottis, pituitary dysplasia and hypopituitarism(2). Congenital heart defects are also not uncommonly found in PHS patients. The diagnosis of PHS is made clinically and radiologically, but can be confirmed genetically by discovery of a mutation in the transcription gene GLI3.

PHS may present in the neonatal period as a complication of the hypothalamic hamartoma as pituitary dysfunction needing resuscitation from adrenal insufficiency, or it may present as respiratory compromise from bifid epiglottis as stridor, feeding difficulties, and recurrent aspiration which may lead to a diagnosis of failure to thrive. During laryngoscopy, the two halves of a bifid epiglottis (BE) may fail to be suspended by the blade and can obscure the glottic inlet. The presence of a BE is fairly rare as demonstrated by the literature review by Urben and Baugh(3) in 1999 which revealed only 20 case reports of BE. In 2000, Ondrey and colleagues (4) studied 27 patients with PHS, 16 of which had a BE (59%). This study identified almost as many patients with a BE as had ever been previously reported in all of the literature. Thus, if an anesthesiologist discovers a BE in a patient with polydactyly, an MRI should be ordered to confirm the diagnosis of Pallister-Hall Syndrome and the patient and family receive genetic counseling.
References:
2. Biesecker L., Gene Reviews, 2006
3. Urben S., Baugh R., Otolaryngology Head and Neck Surgery 1999
4. Ondrey F. et al., American Journal of Medical Genetics 2000