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Mycobacterial Diseases

Chiari I Malformation with Klippel-Trenaunay syndrome: Case report and review of the literature

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Abstract

We present a rare case of an eight-year-old male with Klippel-Trenaunay syndrome (KTS) and a Chiari I malformation (CIM). Magnetic resonance imaging (MRI) to investigate facial asymmetry and speech delay at age two revealed CIM with cerebellar tonsils 1.3 centimeters below the foramen magnum without syringomyelia. The patient underwent a craniectomy and posterior fossa decompression with C1 laminectomy. While gene sequencing determined the patient was negative for the PIK3CA gene mutation, the patient's clinical history strongly suggests KTS. He has hemihypertrophy, leg length discrepancy, hemangiomas and pigmentary mosaicism along the upper and lower extremities, heart murmur, chronic low heart rate, recurrent hip pain, and mild scoliosis. Neurodevelopmental concerns include difficulty reading, attention deficit hyperactivity disorder (ADHD), anxiety, and difficulty running and going downstairs. His most recent MRI shows good decompression at the cervicomedullary junction, global cerebrospinal fluid (CSF) flow, and less peg-like cerebellar tonsils. Also noted were two intravertebral hemangiomas at T5 and T6. While the patient's speech has improved, there is still difficulty with the expressive language. He still has mild delays, runs slowly, and does not alternate feet when climbing stairs. The patient is being followed by multiple specialists including neurology, hematology-oncology, genetics, orthopedic surgery and developmental pediatrics.

Biography

Isabel Snee is a current undergraduate student at the University of Notre Dame. She co-authored this paper during her junior-year intership at the New Jersey Pediatric Neuroscience Institute (NJPNI). She has published three case reports, of which she is primary author, and one literature review.

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