CR45 Hereditary neuropathy with liability to pressure palsies as a possible predisposing factor to the development of Chronic inflammatory demyelinating polyneuropathy

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KEYWORDS: CIDP; PMP22 protein, human; Tomaculous neuropathy

INTRODUCTION/OBJECTIVES: Chronic inflammatory demyelinating polyneuropathy (CIDP) is a symmetric distal muscle weakness and sensory deficit accompanied with diminished or absent deep tendon reflexes. Hereditary neuropathy with liability to pressure palsies (HNPP) is a hereditary neuropathy presenting with transient muscle weakness and sensory symptoms after exposure to pressure or prolonged use in the affected area.
CASE PRESENTATION: A twenty-one-year-old male presented with relapsing-remitting muscle weakness lasting for five years. Symptoms started after exercise and spontaneously regressed after a couple weeks. Physical exam findings included neurological symptoms - paresis of foot dorsiflexion, loss of vibration and hypesthesia, and bilateral m. quadriceps femoris reflex clonus. The patient had a history of essential tremor. Diagnostic tests showed albuminocytological dissociation in the CSF and positive antiganglioside antibodies in the sera. An MR of the L-S spine showed no signs of demyelination. EMG and NCS showed signs of a demyelinating sensory and motor polyneuropathy affecting upper and lower extremities. Genetic testing found a heterozygous deletion of the PMP22 gene, typical for HNPP. The patient was diagnosed with HNPP with superimposed CIDP. Intravenous immunoglobulins were administered at 2 g/kg which improved motor and sensory symptoms. 2 years from the hospital stay, after getting a computer job, the patient reports tingling and weakness in the hands after work, partially regressing during the night.
CONCLUSION: CIDP associated with HNPP has seldom been reported and described in literature. These rare cases outline the possible connection of hereditary neuropathies predisposing to an immune mediated acquired neuropathy.

CR46 Herlyn-Werner-Wunderlich syndrome in a adolescent girl

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KEYWORDS: Hysterectomy; Laparoscopy; Menstrual Cycle; Renal Adysplasia; Uterine Anomalies

INTRODUCTION/OBJECTIVES: Herlyn-Werner-Wunderlich syndrome (HWWS) is a rare congenital genitourinary anomaly with uterine anomalies, unilateral cervicovaginal obstruction, and ipsilateral renal anomalies resulting from the embryological arrest of Müllerian and mesonephric ducts. The onset of nonspecific symptoms occurs after menarche. Having two normal uteri and obstructed cervix or hemivagina, the patient will have regular menses through the non-obstructed vaginal side, coinciding with cyclic pelvic pain from the encumbered blood in the obstructed side. The diagnosis is often delayed until acute complications develop.
CASE PRESENTATION: A 12-year-old girl presented to the emergency department with LLQ pain that started on the third day of her menstrual cycle. She had menarche a year ago and has no history of dysmenorrhoea. Vital signs, laboratory parameters, abdominal ultrasound, and RTG were unremarkable. On the fifth day of the menstrual cycle, with symptoms of persistent, now RLQ pain, elevated body temperature, and peritoneal defense, the patient was diagnosed with acute appendicitis. Intraoperative examination revealed an innocent appendix, bloody secretion in the pelvis, and an oedematous left fallopian tube. HWWS diagnosis was established after an emergency consultation with a gynecologist during the operation. MRI provided more detailed information for surgical strategies, which confirmed the diagnosis: uterus didelphys with left cervical atresia enlarged left uterine horn, left hematosalpinx, possible hemivagina, and left kidney agenesis. Laparoscopic left-side hysterectomy and salpingectomy were performed.
CONCLUSION: HWWS should be suspected in cases with cyclic pelvic pain and renal malformations. This case highlights the importance of knowing developmental anomalies to include them in the differential diagnosis.