

Prader-Willi Syndrome: Clinical Concerns for the Orthopedic Surgeon

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Introduction: Prader-Willi Syndrome (PWS) is a chromosome 15 disorder characterized by hypotonia, hypogonadism, hyperphagia and obesity. Musculoskeletal manifestations, including scoliosis, hip dysplasia and lower limb alignment abnormalities, are well described in the orthopedic literature. However, care of this patient population from the orthopedic surgeon's perspective is complicated by other clinical manifestations of PWS. Osteopenia, psychiatric disorders, and diminished pain sensitivity are frequently noted in PWS but are not discussed in the orthopedic literature. The authors present a clinical review of an 8-year experience caring for 31 patients with PWS to highlight all clinical concerns that influence orthopedic management.



Methods: Thirty-one institutionalized patients diagnosed with PWS were examined and all past medical records were reviewed. Patient demographics, genetic testing, musculoskeletal diagnoses, psychiatric diagnoses, and clinical behaviors were recorded. Radiological studies performed in the course of routine clinical care were evaluated.

Results: Twenty-two men and 9 women, average age 22 years (range 8-39 years), were studied. A chromosome 15Q abnormality was confirmed in 18 patients. Scoliosis was clinically detected in 24 of 31 patients and confirmed by radiographs in 14 of these 24 patients (45% overall with scoliosis) with an average primary curve of 31°; three were braced and 2 underwent spinal fusion. Radiographs also revealed diminished cervical lordosis and increased cervicothoracic kyphosis in 16 patients, a previously undescribed finding. Hip radiographs of 26 patients revealed dysplasia in 2 patients (13%); no SCFEs were identified. Fourteen patients had sustained a total of 58 fractures with 6 patients sustaining multiple fractures (range 2-7). Bone densitometry was performed on 14 patients; 8 patients had osteopenia and 4 had osteoporosis based on lumbar spine Z-scores. Twenty-six patients had axis I psychiatric diagnoses including impulse control disorder (7), organic personality disorder (6), oppositional defiant disorder (5), dysthymic disorder (4), depressive disorder NOS (3), ADHD (2), and OCD (2). Nine patients exhibited self-mutilating behaviors. Six patients have undergone orthopedic surgical procedures with 1 major complication (spinal infection). Fracture management was associated with frequent minor complications.

Discussion: Osteopenia, poor impulse control and defiant behaviors, and diminished pain sensitivity are aspects of PWS that may complicate all facets of orthopedic non-surgical and surgical management in this patient population. The treating orthopedic surgeon must plan carefully and proceed with caution when treating children and adults with PWS.

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