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Overview on Hersh Podruch Weisskopk Syndrome

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Perspective

IBMPFD (inclusion body myopathy with Paget disease of bone and frontotemporal dementia) is a multisystem degenerative genetic disorder characterised by adult-onset proximal and distal muscle weakness (clinically resembling limb-girdle muscular dystrophy; see this term); early-onset Paget disease of bone (see this term), manifesting with bone pain, deformity, and enlargement of the long bones; and premature frontotemporal dementia (see this term Muscle weakness affects the other limbs and respiratory muscles as the condition advances, eventually leading to respiratory or cardiac failure. IBMPFD (inclusion body myopathy with Paget disease of bone and frontotemporal dementia) is a multisystem degenerative genetic disorder characterised by adult-onset proximal and distal muscle weakness (clinically similar to limb-girdle muscular dystrophy; see this term); early-onset Paget disease of bone (see this term), manifesting with bone pain, deformity, and enlargement of the long bones; and premature frontotemporal dementia (see this term), manifesting Muscle weakness in the other limbs and respiratory muscles worsens as the condition advances, eventually leading to respiratory or cardiac failure.

Early-onset inclusion body myopathy the muscles, bones, and brain can all be affected by Paget disease and frontotemporal dementia (IBMPFD). Muscle weakness (myopathy) is frequently the first sign of IBMPFD, which arises in mid-adulthood. The hips and shoulders are the first muscles to become weak, making it difficult to ascend stairs and raise the arms over the shoulders. Other muscles in the arms and legs become weak as the illness progresses. Muscle weakness can impact the respiratory and cardiac muscles, resulting in life-threatening breathing problems and heart failure. A illness called Paget disease of bone affects around half of all people with IBMPFD. The hips, spine, and skull, as well as the long bones of the arms and legs, are the most commonly affected bones in this illness.

The most common symptom of Paget disease is bone pain, especially in the hips and spine. This syndrome can occasionally cause bones to become so weak that they break (fracture). The condition affects the brain in around one-third of patients with IBMPFD. IBMPFD is linked to frontotemporal dementia, a type of dementia that appears in people in their forties and fifties. Frontotemporal dementia patients may have difficulty communicating, recalling words and names (dysnomia), and utilising numbers at first (dyscalculia). Parts of the brain that control logic, personality, social skills, speech, and language are damaged over time by the disorder. The condition is also marked by personality changes, a loss of judgement, and improper social behaviour. As dementia progresses, those who are affected become unable to speak, read, or care for themselves.

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