Inclusion Body Myopathy with Early-Onset Paget Disease with or without Frontotemporal Dementia 1

National Cancer Institute

Source

National Cancer Institute. Inclusion Body Myopathy with Early-Onset Paget Disease with or without Frontotemporal Dementia 1. NCI Thesaurus. Code C122663.

A rare autosomal dominant inherited disorder caused by mutations in the VCP gene. It can affect the muscles, bones, and brain. Patients may develop myopathy that initially involves the muscles of the hips and shoulders and as the disorder progresses it may affect the cardiac and respiratory muscles, leading to life-threatening cardiac and pulmonary failure. Approximately half of the adults develop Paget disease of bone, and approximately one-third develop frontotemporal dementia.