Spondyloepimetaphyseal Dysplasia with Joint Laxity Type 2

National Cancer Institute

Source

National Cancer Institute. Spondyloepimetaphyseal Dysplasia with Joint Laxity Type 2. NCI Thesaurus. Code C125419.

A rare disorder caused by mutation in the KIF22 gene. It is characterized by short stature, midface retrusion, progressive knee malalignment, generalized ligamentous laxity, and mild spinal deformity.