Wormian bone-multiple fractures-dentinogenesis imperfecta-skeletal dysplasia

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Wormian bone-multiple fractures-dentinogenesis imperfecta-skeletal dysplasia. ORPHA:166277

Skeletal dysplasia with wormian bone-multiple fractures-dentinogenesis imperfecta is a skeletal disorder, reported in three patients to date, characterized clinically by multiple fractures, wormian bones of the skull, dentinogenesis imperfecta and facial dysmorphism (hypertelorism, periorbital fullness). Although the signs are very similar to osteogenesis imperfecta, characteristic cortical defects in the absence of osteopenia and collagen abnormalities are considered to be distinctive. There have been no further descriptions in the literature since 1999.