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Aspek klinik, genetik dan molekuler osteogenesis imperfekta

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Abstrak

Osteogenesis imperfecta (OI) is a heritable disorder of connective tissue that mainly affects the bones. Being always associated with bone fragility, it is also known as "brittle bone" disease. Multiple bone fractures are common, and in severe cases can occur even before birth. The condition is characterized by fractures with minimal or absent trauma, dentinogenesis impercta, short stature, blue sclerae, and in adult years, hearing loss. Most cases of OI, which is inherited in an autosomal dominant manner, result from mutations affecting the genes COLIA1 (collagen type 1 alpha 1) and COLIA2 (collagen type 1 alpha 2) that encode pro-a 1 and pro-a 2 chains of type 1 collagen. The type 1 collagen molecule accounts for about 90% of the organic matrix of the bone. In addition, collagen forms a family of proteins that strengthen and support many tissues in the body, including cartilage, tendons, skin, and the white part of the eye (sclera). This paper aims to review the genetic contribution to OI.