Table 1. Human sclerostin deficiency

Disease	Sclerosteosis	Van Buchem disease
Genetic defect	Loss-of-function mutation of SOST gene on chromosome 17q12-q21	52 kb deletion downstream of SOST
Prevalence	High in Afrikaners of South Africa	High in Urk in the Netherlands
Homozygotes	• undetectable serum sclerostin levels • dense bones • tall stature • syndactyly	• low serum sclerostin level • phenotype similar to sclerosteosis but less severe • no tall stature • no syndactyly
Heterozygotes	• phenotypically normal • low but detectable serum sclerostin levels • dense bones • rarely fracture	• phenotypically normal