

Orthopaedic Aspect of Type II Collagenopathy

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Type II collagenopathy has a wide variety of phenotype. A series of COL2A1 mutation-confirmed patients were reviewed for orthopaedic aspects and their surgical intervention. Forty-two patients from 39 families were the subjects of this review. Their clinical diagnosis included SEDC (17), late onset SED (8), Kniest dysplasia (4), Stickler syndrome (3), SED Stanescu type (3), SED with early-onset scoliosis (3), spondyloperipheral dysplasia (2), Czech dysplasia (1), and bilateral avascular necrosis of the hip (1). Short stature ($z < -2$) was observed in 64.3%. Height with $z > -2.0$ was observed in Czech dysplasia, late onset SED, SED Stanescu type etc. Although most of the patients showed odontoid hypoplasia, C1-2 instability was confirmed by C-spine flexion-extension radiographs in 33%. Significant scoliosis was observed in 14.3%. Hip joint frequently showed clinical and radiographic abnormalities. Delayed and abnormal ossification and femoral

head deformity was common. Avascular necrosis of the femoral head (AVN) is sometimes difficult to differentiate from abnormal ossification process. Sequential radiographic changes of AVN along with compatible clinical finding were observed only in five cases.

Twenty-four orthopaedic surgeries were performed. Seventeen (70.8%) hip surgeries were performed including containment surgeries on the femoral side (3) or acetabular side (5); redirection osteotomies (9) for coxa vara, deformed or dislocated femoral head. Genu valgum correction was done in three cases. C1-2 (1) or thoracolumbar (2) spine was stabilized by posterior fusion in three cases. One patient of SED with early-onset scoliosis showed unilateral tibial undergrowth, and underwent tibial lengthening procedure, exceptionally.

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