Recessive multiple epiphyseal dysplasia (rMED) with homozygosity for C653S mutation in the DTDST gene -phenotype, molecular diagnosis and surgical treatment of habitual dislocation of multilayered patella: case report

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Multiple epiphyseal dysplasia (MED) is one of the more common generalised skeletal dysplasias. Due to its clinical heterogeneity diagnosis may be difficult. Mutations of at least six separate genes can cause MED. Joint deformities, joint pain and gait disorders are common symptoms.; We report on a 27-year-old male patient suffering from clinical symptoms of autosomal recessive MED with habitual dislocation of a multilayered patella on both sides, on the surgical treatment and on short-term clinical outcome. Clinical findings were: bilateral hip and knee pain, instability of femorotibial and patellofemoral joints with habitual patella dislocation on both sides, contractures of hip, elbow and second metacarpophalangeal joints. Main radiographic findings were: bilateral dislocated multilayered patella, dysplastic medial tibial plateaus, deformity of both femoral heads and osteoarthritis of the hip joints, and deformity of both radial heads. In the molecular genetic analysis, the DTDST mutation g.1984T >A (p.C653S) was found at the homozygote state. Carrier status was confirmed in the DNA of the patient's parents. The mutation could be considered to be the reason for the patient's disease. Surgical treatment of habitual patella dislocation with medialisation of the tibial tuberosity led to an excellent clinical outcome.; The knowledge of different phenotypes of skeletal dysplasias helps to select genes for genetic analysis. Compared to other DTDST mutations, this is a rather mild phenotype. Molecular diagnosis is important for genetic counselling and for an accurate prognosis. Even in case of a multilayered patella in MED, habitual patella dislocation could be managed successfully by medialisation of the tibial tuberosity.
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