fibrodysplasia ossificans progressiva

I read with interest the article by Connor et al. on the clinical and pathologic spectrum of fibrodysplasia ossificans progressiva (J Med Genet 1993;30:867-9). The article describes the clinical and pathologic features of this disorder, including the typical presentation of progressive ossification of skeletal muscle, tendons, and fascia, as well as the associated skeletal abnormalities and cardiovascular anomalies. The authors also discuss the genetic basis of the disorder, which is caused by mutations in the fibroblast growth factor receptor 4 gene (FGFR4). The prevalence of the disorder is estimated to be 1 in 100,000 live births, and the incidence is higher in males than in females.

Low segregation ratios in autosomal recessive disorders

I read with interest the article by Bondy and Young on the segregation of autosomal recessive traits in families (J Med Genet 1993;30:621-4). The authors reviewed 111 autosomal recessive disorders and found that the segregation ratio was significantly lower than 1:1 in only 5 cases. They hypothesize that this may be due to the presence of dominant negative mutations or other genetic or environmental factors that influence the clinical expression of the disorder.

Cutis laxa: a feature of Costello syndrome

We were extremely interested to read the letter 'Cutis laxa and the Costello syndrome' by Agnese et al., which described the occurrence of cutis laxa in three siblings with Costello syndrome. Cutis laxa is a rare hereditary condition characterized by skin elasticity and joint hyperlaxity, and is caused by mutations in the COL5A1 gene. The association of cutis laxa with Costello syndrome suggests a common genetic basis for these two conditions, and may provide insights into the pathogenesis of cutis laxa.

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