

DMC and SMS syndromes are rare genetic disorders. As of 2007 there were over 90 individuals with DMC or SMS reported in the literature (Lachman, 2007, p 934) who were from a number of different ethnic groups (El Ghouzzi et al. 2003; Pogue et al. 2005; Aglan et al. 2009). A diagnosis of DMC syndrome may be suspected upon a thorough clinical evaluation, a detailed patient history, and identification of characteristic clinical findings, e.g., barrel chest and disproportionate short stature. Radiographs may confirm specific skeletal abnormalities and findings consistent with DMC syndrome and includes notching of the vertebral bodies, lacy appearance of the iliac crest, and small and malformed carpal bones. Alternatively, gene testing for mutations in DYM can be done.