

Polycystic kidney disease in a patient with achondroplasia: case report.

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Abstract

Autosomal dominant polycystic kidney disease is a multisystem disease involving many organs. An association with other diseases such as tuberous sclerosis, von Hippel-Lindau disease and Marfan syndrome have been previously described. We describe a 35 year old female with achondroplasia who developed polycystic kidney disease involving both kidneys and progressing to end-stage renal disease. To the best of our knowledge this is the first such case described in the literature. We also delve, briefly, into the possibility of the genes and chromosomes involved in Marfan syndrome, polycystic kidney disease, tuberous sclerosis and achondroplasia playing a role in the co-occurrence of these entities