

Familial pattern and inheritance of Dupuytren's disease: a comparison of populations and pedigrees in the UK and Iceland

Hindocha S, Gudmundsson K, Arngrimson R, Jonsson T, McGrouther DA, Bayat A

Introduction

Dupuytren's disease (DD) is a familial disorder of unknown aetiology. Understanding the exact inheritance pattern in DD is of relevance for better understanding and clinical management of the disease. A variety of inheritance patterns with a variable penetrance have been reported to date. The aim of this study was to compare the inheritance patterns of DD in two different populations which are commonly affected with DD.

Method

Patients diagnosed with DD ($n = 135$) were randomly selected from two hospitals in Manchester, and Wrightington in the UK as well as in Reykjavik, Iceland. Family pedigrees were drawn for each patient with a positive family history and analysed. Demographic details, environmental risks, and the severity of disease were assessed. The variant penetrance in the UK DD cohort was estimated by calculating the population attributable risk.

Results & conclusion

The presence of DD in 979 relatives was obtained from 60 (44%) patients with a positive family history. Patients with a family history had a greater severity of disease than those who did not ($p < 0.05$). An autosomal dominant pattern of inheritance with full penetrance was identified in 38 (63%) cases. The level of penetrance in the whole DD cohort was estimated at 35%. The population attributable risk was calculated at 22%. A unique Icelandic family with 22 affected members in a family of 45 members showed autosomal dominance with complete penetrance. We have identified a pattern of inheritance in familial DD which can be used to calculate the relative risk of this condition. This can be used in clinical practice to counsel patients on the likelihood of their family members developing DD in addition to planning the correct surgical treatment. Those with a stronger family history and therefore more severe disease may benefit from dermofasciectomy as a primary treatment.