Int J Hum Genet, 1(2): 133-137 (2001)
PRINT: ISSN 0972-3757 ONLINE: ISSN 2456-6330
DOI: 10.31901/24566330.2001/01.02.11

Clinical Genetic Analysis of Retinitis Pigmentosa in Indian Population

Subhabrata Chakrabarti[a], Virinder Kaur Sarhadi[a], Daljit Singh[b], Indu Ravijit Singh[b] and Jai Rup Singh[a]*

[a]Centre for Genetic Disorders, Guru Nanak Dev University, Amritsar, India;
[b]Dr. Daljit Singh Eye Hospital, Amritsar, India

KEY WORDS Pigmentary retinopathy; Retinitis pigmen-tosa; Retinal degeneration; Segregation

ABSTRACT Four hundred families affected with Retinitis Pigmentosa (RP), from 16 different states of India were analysed to ascertain segregation pattern of RP and to study the association of clinical variables like “age of onset” and “rate of progression” with the different clinical subtypes of RP. Families were categorized on the basis of clinical types and inheritance pattern. Segregation analysis, based on the maximum likelihood estimate, was carried out on 426 sibships from 400 families. 75% of the cases were autosomal recessive (AR), 10% autosomal dominant (AD) and 1% X-linked. In the remaining 14%, the inheritance pattern was not clear. Segregation analysis of the sibship data showed good agreement with respect to the AR cases, with a segregation value of 0.23. Penetrance was relatively low (42%) in AD cases. Segregation analysis of AR cases, with the inclusion of simplex cases, indicated that these cases might be having an underlying recessive mode of inheritance. High frequency of recessive cases could be attributed to inbreeding and consanguinity in families. Statistically significant differences with respect to “age of onset” and “rate of progression” were observed among different clinical types of RP, which could be of importance in counseling.

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