Clinical and genetic features in autosomal recessive and X-linked Alport syndrome


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Abstract

Background: This study determined the family history and clinical features that suggested autosomal recessive rather than X-linked Alport syndrome. Methods: All patients had the diagnosis of Alport syndrome and the mode of inheritance confirmed by genetic testing, and underwent examination at a single centre. Results: Patients comprised 9 males and 6 females with autosomal recessive Alport syndrome, and 15 males and 22 females with X-linked disease. Fourteen (93%) individuals with autosomal recessive Alport syndrome developed early-stage renal failure, all 15 had hearing loss, and most had lenticonus (12, 80%), and a central (13, 87%) or peripheral (13, 87%) retinopathy. These features occurred as often as in males with X-linked disease. Females with autosomal recessive inheritance were less likely to have an affected family member in another generation (p = 0.01) than females with X-linked disease. They were more likely to have renal failure (p = 0.003), hearing loss (p = 0.02) and lenticonus (p < 0.001). Fifty percent had a central retinopathy compared with 18% with X-linked disease (p = 0.14), but peripheral retinopathy prevalence was not different (p = 0.64). Nonsense mutations accounted for 67% (8/12) of these disease-causing mutations. Conclusions: Autosomal recessive inheritance is increased in females with Alport syndrome and early onset renal failure, hearing loss, lenticonus, and, possibly, central retinopathy. © 2013 IPNA.

Author keywords

Alport syndrome, Nonsense mutations, Retinopathy.

Indexed keywords

MeSH:

Adolescent Adult DNA Mutational Analysis Female Genes, X-Linked Genetic Predisposition to Disease Genetic Testing Hearing Loss Heredity Humans Kidney Failure, Chronic Male Middle Aged Mutation Nephritis, Hereditary Pedigree Phenotype Predictive Value of Tests Prognosis Retinal Diseases Risk Factors Sex Factors Time Factors Young Adult.