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

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Abstract

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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 18 males and 22 females with X-linked disease. Fourteen (93 %) individuals with autosomal recessive Alport syndrome developed early end-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

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