Relative frequencies of inherited retinal dystrophies and optic neuropathies in Southern France: assessment of 21-year data management

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Résumé en anglais	**PURPOSE**: Inherited retinal dystrophies (IRDs) and inherited optic neuropathies (IONs) are rare diseases defined by specific clinical and molecular features. The relative prevalence of these conditions was determined in Southern France. **METHODS**: Patients recruited from a specialized outpatient clinic over a 21-year period underwent extensive clinical investigations and 107 genes were screened by polymerase chain reaction/sequencing. **RESULTS**: There were 1957 IRD cases (1481 families) distributed in 70% of pigmentary retinopathy cases (56% non-syndromic, 14% syndromic), 20% maculopathies and 7% stationary conditions. Patients with retinitis pigmentosa were the most frequent (47%) followed by Usher syndrome (10.8%). Among non-syndromic pigmentary retinopathy patients, 84% had rod-cone dystrophy, 8% cone-rod dystrophy and 5% Leber congenital amaurosis. Macular dystrophies were encountered in 398 cases (30% had Stargardt disease and 11% had Best disease). There were 184 ION cases (127 families) distributed in 51% with dominant optic neuropathies, 33% with recessive/sporadic forms and 16% with Leber hereditary optic neuropathy. Positive molecular results were obtained in 417/609 families with IRDs (68.5%) and in 27/58 with IONs (46.5%). The sequencing of 5 genes (ABCA4, USH2A, MYO7A, RPGR and PRPH2) provided a positive molecular result in 48% of 417 families with IRDs. Except for autosomal retinitis pigmentosa, in which less than half the families had positive molecular results, about 75% of families with other forms of retinal conditions had a positive molecular diagnosis. **CONCLUSIONS**: Although gene discovery considerably improved molecular diagnosis in many subgroups of IRDs and IONs, retinitis pigmentosa, accounting for almost half of IRDs, remains only partly molecularly defined.
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