

RISCALW a windows program for risk calculation in Duchenne muscular dystrophy.

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In many DMD families risk calculation is still an important part of genetic counseling particularly if the patient is not available or a mutation is not detectable.

A prerequisite for risk calculation in families is an appropriate genetic model for the disease of interest. Because of empirical evidence that the formerly used simple genetic model for Duchenne muscular dystrophy (DMD) is not sufficient, a general model to include germline mosaicism and heterogeneous new mutation rates depending on sex and mutation type has been formulated. Based on this model RISCALW was developed. RISCALW is a user-friendly Windows program for risk calculation in families with DMD. It runs on any standard personal computer under Windows operating systems.

A range of different genetic models including germline mosaicism, sex and mutation specific mutation rates may be used. It is possible to include marker genotypes, creatininkinase values and FISH-deletion test results. Pedigree data input is required in a modified LINKAGE format in the pedigree window; the user can make selections for the risk calculation in the parameter window; the results are displayed in the log window. The main editing features are accessible for pedigrees and results. Pedigrees can be included via the Windows clipboard from any wordprocessor. Pedigrees and results can be added to any wordprocessor in the same manner. Detailed online help is available. RISCALW has been extensively tested for more than 100 families. The user is able to perform risk calculation in large families in an easy way and thus explore the dependence of results on different model assumptions for example different mutation rates in male and female.

RISCALW is freely available from the authors for scientific use.

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