RISCALW a windows program for risk calculation in Duchenne muscular dystrophy

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Calculation with paper and pencil may be error prone
Principle features of RISCALW

pedigree window

parameter window

output window
Possibilities for the genetic model
FISH test and CK measurements

CK as odds ratios

CK quantitatively
Marker characteristics

<table>
<thead>
<tr>
<th>Nr.</th>
<th>Name</th>
<th>Use</th>
<th>Alleles</th>
<th>Allel Frequencies</th>
</tr>
</thead>
<tbody>
<tr>
<td>1</td>
<td>M1</td>
<td>✓</td>
<td>1</td>
<td>1</td>
</tr>
<tr>
<td>2</td>
<td>M2</td>
<td>✓</td>
<td>2</td>
<td>0.5 : 0.5</td>
</tr>
<tr>
<td>3</td>
<td>M3</td>
<td>✓</td>
<td>3</td>
<td>0.3333 : 0.3333 : 0.3333</td>
</tr>
</tbody>
</table>
Marker map

a

b

c
Choice of counselee and calculation start

Carrier probabilities for Son of 6:
- Mutation (PM+Del) = 4.5680%
- Deletion = 0.6185%
- Mosaicism (PM+Del) = 0.0059%
- Del-mosaicism = 0.0014%
Technical details

- RISCALW is programmed in Delphi
- runs on PC under any WIN.. operation system
- uses a new model to account for germline mosaicism and sex specific mutation rates
Program validation

- extensively tested
- compared with LINKAGE
- compared with hand calculations
Concluding remarks

- easy to use risk calculation for DMD
- flexible
- only program which is able to account for mosaicism, heterogeneous new mutation rates
- incorporation of negative deletion tests, CK-values and marker genotypes possible
- sensitivity analysis very convenient