

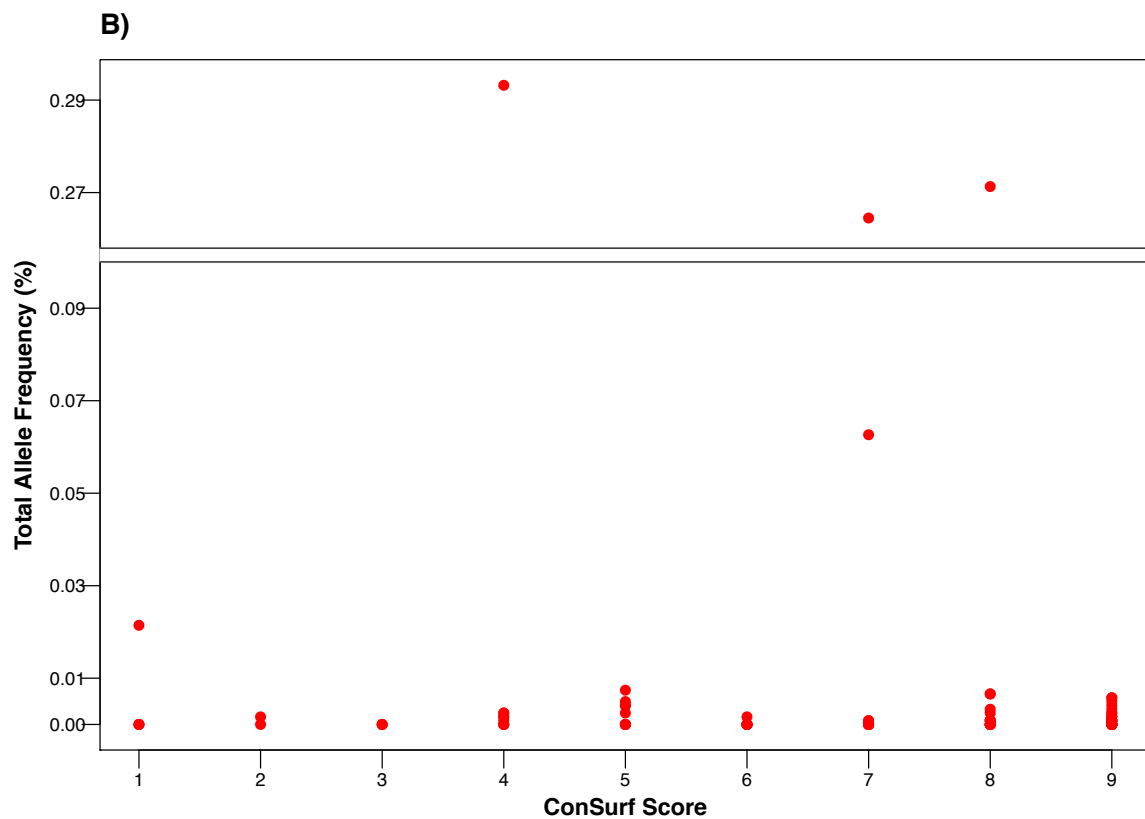
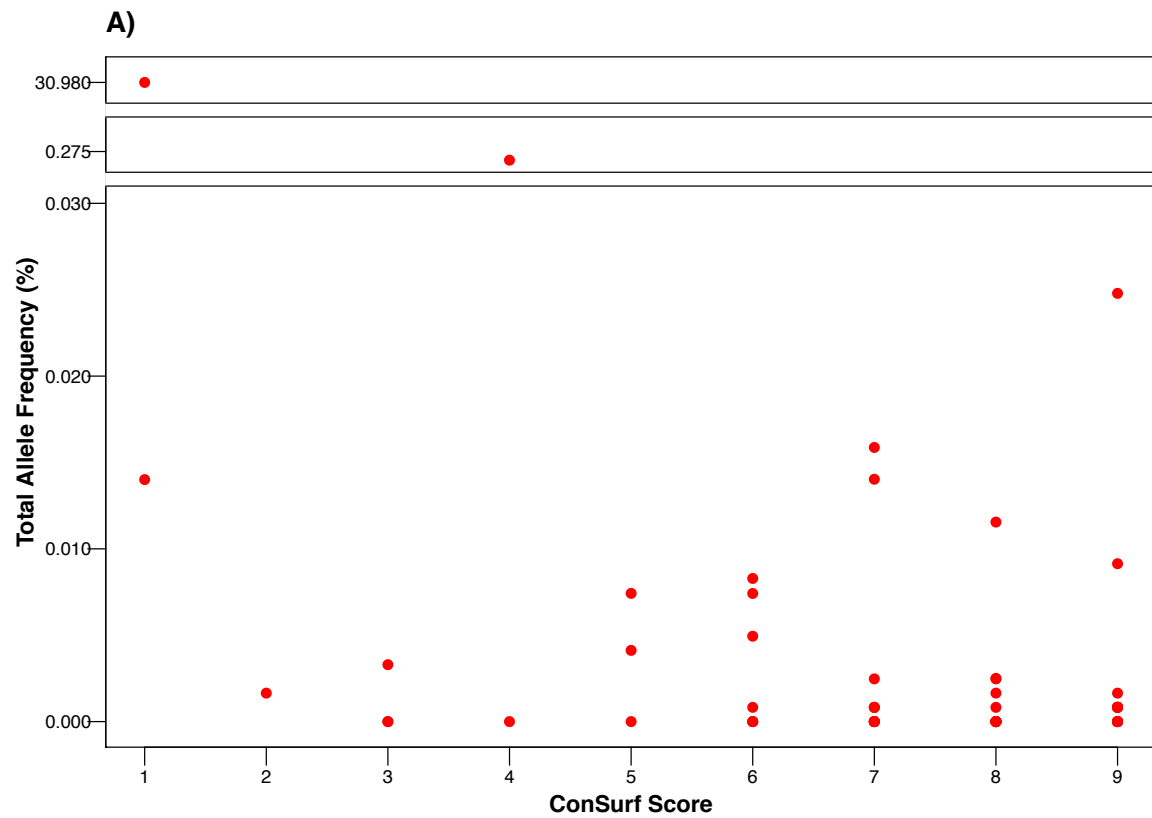


Figure S1. Comparison of allele frequency (based on ExAC) and evolutionary conservation (based on ConSurf scores). A) Cystinuria associated mutations of b(0+)AT. B) Cystinuria associated mutations of rBAT.

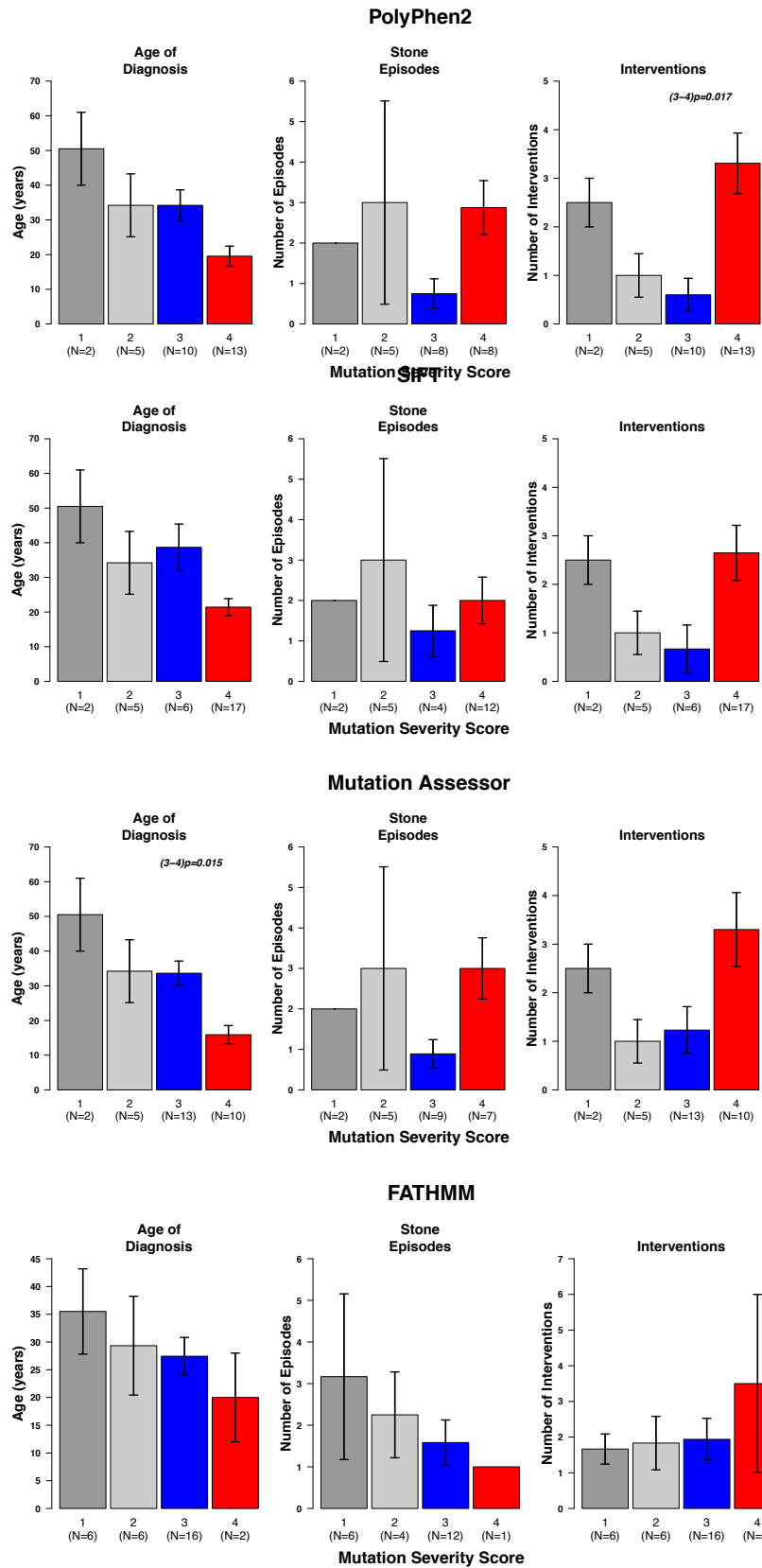


Figure S2. Comparison of other clinical parameters between the different severity score groups, for individuals with b(o+)AT mutations. There is one plot per prediction method (PolyPhen2, SIFT Mutation Assessor, and FATHMM). The group numbers are given at the bottom of the plots, with the sample number given in brackets underneath the group name. Where significant differences between groups occur ($p < 0.05$) the p-value is displayed on the plot, e.g. (1-2) $p = 0.001$ means a significant difference between groups 1 and 2.

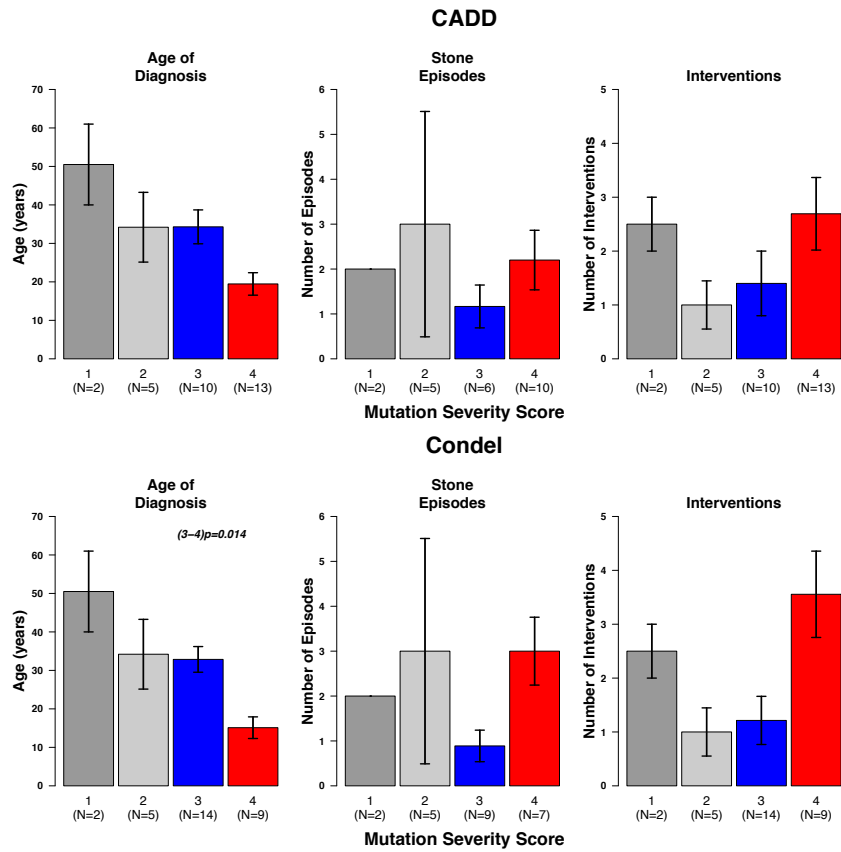


Figure S3. Comparison of other clinical parameters between the different severity score groups, for individuals with b(o+)AT mutations. There is one plot per integrated prediction method (CADD and Condel). The group numbers are given at the bottom of the plots, with the sample number given in brackets underneath the group name. Where significant differences between groups occur ($p < 0.05$) the p-value is displayed on the plot, e.g. (1-2) $p=0.001$ means a significant difference between groups 1 and 2.

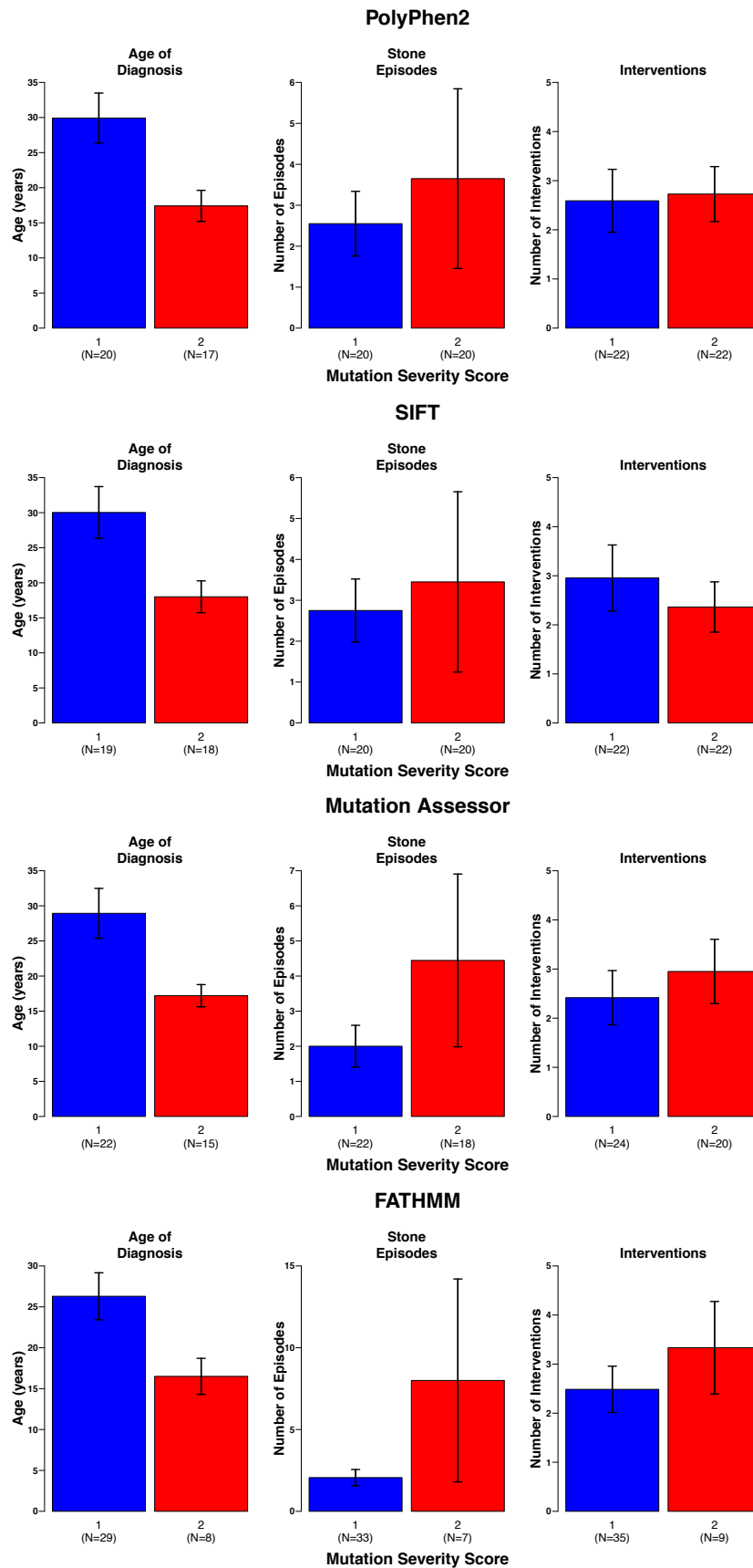


Figure S4. Comparison of other clinical parameters between the different severity score groups, for individuals with rBAT mutations. There is one plot per prediction method (PolyPhen2, SIFT Mutation Assessor, and FATHMM). The group numbers are given at the bottom of the plots, with the sample number given in brackets underneath the group name. Where significant differences between groups occur ($p < 0.05$) the p-value is displayed on the plot, e.g. (1-2) $p=0.001$ means a significant difference between groups 1 and 2.

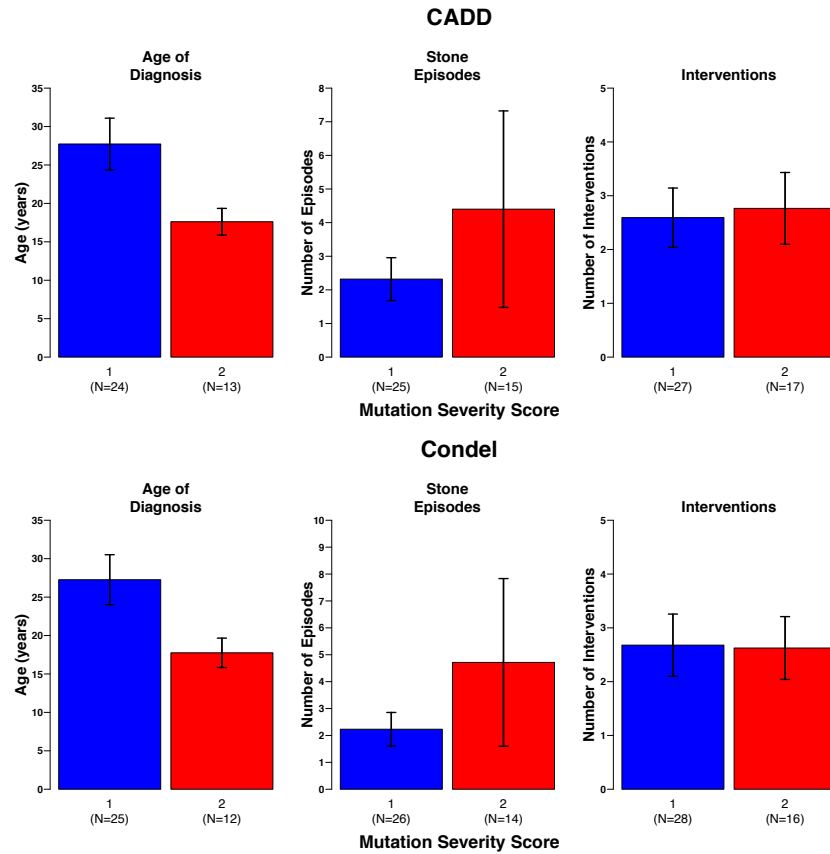


Figure S5. Comparison of other clinical parameters between the different severity score groups, for individuals with rBAT mutations. There is one plot per integrated prediction method (CADD and Condel). The group numbers are given at the bottom of the plots, with the sample number given in brackets underneath the group name. Where significant differences between groups occur ($p < 0.05$) the p-value is displayed on the plot, e.g. (1-2) $p = 0.001$ means a significant difference between groups 1 and 2.