**Supplementary Table 1:** Molecular information.

| ID | Patient # | *CHM* Mutation | Type of Mutation | Exon/Intron Location | Predicted Change |
|---|---|---|---|---|---|
| 12001 | | c.519delAG** | frameshift | Exon 5 | p.? |
| 13004 | P6 | deletion of exon 1 | deletion | Exon 1 | REP1 absent |
| 13005 | | c.1663A>T | nonsense | Exon 14 | p.Arg555* |
| 13012 | | c.189+1 G>T | splice site | Intron 3 | p.? |
| 13013 | | NA | NA | NA | NA |
| 13015 | P5 | del. entire gene | deletion | del. entire gene | REP1 absent |
| 13017 | | C | C | C | C |
| 13019 | | NA | NA | NA | NA |
| 13020 | | NA | NA | NA | NA |
| 13027 | | c.1185C>G | nonsense | Exon 9 | p.Ser395* |
| 13031 | P2 | deletion exons 3-8** | deletion | Exons 3-8 | p.? |
| 13032 | | c.757C>T | nonsense | Exon 6 | p.Arg253* |
| 13034 | | c.838C>T | nonsense | Exon 6 | p.Arg270* |
| 13035 | | NA | NA | NA | NA |
| 13039 | P4 | c.1327_1328delAT | frameshift | Exon 10 | p.? |
| 13040 | | deletion exons 2-4 | deletion | Exons 2-4 | p,? |
| 13044 | P19 | c.315_318delTCAG | frameshift | Exon 5 | p.? |
| 13045 | | NA | NA | NA | NA |
| 13048 | | c.700A>T | nonsense | Exon 5 | p.Lys234* |
| 13050 | | c.700A>T | nonsense | Exon 5 | p.Lys234* |
| 13051 | | c.799C>T | nonsense | Exon 6 | p.Arg267* |
| 13053 | | c.281delA** | frameshift | Exon 3 | p.? |
| 13055 | | c.838C>T | nonsense | Exon 6 | p.Arg270* |
| 13056 | | c.445delT | frameshift | Exon 5 | p.? |
| 13057 | P13 | c.745delT | nonsense | Exon 6 | p.Arg239* |
| 13061 | | del exon 9,10 | deletion | exons 9, 10 | p.? |
| 13062 | | c.1649 T>C | missense | Exon 14 | p.Leu550Pro |
| 13063 | | c. 1649 T>C | missense | Exon 14 | p.Leu550Pro |
| 13064 | | del exons 1-2 | deletion | Exons 1, 2 | REP1 absent |
| 13066 | | del exons 1-2 | deletion | Exons 1, 2 | REP1 absent |
| 13067 | | c.838C>T | nonsense | Exon 6 | p.Arg270* |
| 13068 | | C | C | C | C |
| 13070 | | NA | NA | NA | NA |
| 13071 | | c.1663A>T | nonsense | Exon 14 | p.Arg555* |
| 13072 | | c.1771-1g>a | splice site | Intron 14 | p.? |
| 13073 | | c.1218C>A | nonsense | Exon 9 | p.Cys406* |
| 13074 | | c.1218C>A | nonsense | Exon 9 | p.Cys406* |
| 13075 | | c.1218C>A | nonsense | Exon 9 | p.Cys406* |
| 13076 | | c.1218C>A | nonsense | Exon 9 | p.Cys406* |
| 13078 | | del exons 1-15 | deletion | Exons 1-15 | REP1 absent |
| 13080 | | c.817_818insC** | frameshift | Exon 6 | p.? |
| 13085 | | del exons 2-4 | deletion | Exons 2-4 | REP1 absent |
| 13086 | | c.1663A>T | nonsense | Exon 14 | p.Arg555* |
| 13087 | | c.1663A>T | nonsense | Exon 14 | Arg555ter |
| 13092 | | c.889A>T | nonsense | Exon 7 | p.Lys297* |
| 13099 | P3 | p.757C>T | nonsense | Exon 6 | p.Arg253* |
| 13100 | | p.757C>T | nonsense | Exon 6 | p.Arg253* |
| 13101 | | c.838C>T | nonsense | Exon 6 | p.Arg270* |
| 13102 | | c.838C>T | nonsense | Exon 6 | p.Arg270* |

| | | | | | |
|---|---|---|---|---|---|
| 13106 | | c.940-1g>t | splice site | Intron 7 | p.? |
| 13117 | | c.820-2a>g | splice site | intron 6 | p.? |
| 13118 | | c.49+5g>t | splice site | intron 1 | p.? |
| 13122 | | c.808C>T | nonsense | Exon 6 | p.Arg270* |
| 13125 | | c.1437dupA | frameshift | Exon 12 | p.? |
| 13126 | | c.580_581delGA** | frameshift | Exon 5 | p.? |
| 13130 | | c.1327_1328delAT | frameshift | exon 10 | p.? |
| 13131 | P9 | .1327_1328delAT | frameshift | exon 10 | p.? |
| 13132 | | .1327_1328delAT | frameshift | exon 10 | p.? |
| 13136 | | c.1520A>G | missense | Exon 13 | p.His507Arg |
| 13144 | | c.715C>T | nonsense | Exon 6 | p.Arg239* |
| 13146 | | NA | NA | NA | NA |
| 13151 | | c.1161C>T | nonsense | exon 9 | p.Gln385* |
| 13159 | | c.1144G>T | nonsense | exon 9 | p.Glu382* |
| 13167 | | del. entire gene | deletion | del. entire gene | REP1 absent |
| 13170 | P17 | c.757C>T | nonsense | Exon 6 | p.Arg253* |
| 13172 | | c.1166+2T>G | splice site | Intron 8 | p.? |
| 13173 | | NA | NA | NA | NA |
| 13177 | | c.519delAG | frameshift | Exon 5 | p.? |
| 13181 | | c.116+1 G>A | splice site | Intron 2 | p.? |
| 13183 | | c.745delT** | frameshift | | p.? |
| 13185 | | c.1631delA | frameshift | Exon 14 | p.? |
| 13186 | P14 | c.1631delA | frameshift | Exon 14 | p.? |
| 13188 | P16 | c.757C>T | nonsense | Exon 6 | p.Arg253* |
| 13190 | P8 | del. entire gene | deletion | del. entire gene | REP1 absent |
| 13193 | | C | C | C | C |
| 13195 | | c.820-2a>g | splice site | intron 6 | p.? |
| 13197 | P7 | deletion exons 3-8 | deletion | Exons 3-8 | p.? |
| 13200 | | c.910G>T | nonsense | exon 7 | p.Glu304* |
| 13203 | P15 | NA | NA | NA | NA |
| 13204 | | NA | NA | NA | NA |
| 13205 | | NA | NA | NA | NA |
| 13208 | | c.519delAG | frameshift | Exon 5 | p.? |
| 13209 | P7 | NA | NA | NA | NA |
| 13210 | | del. entire gene | deletion | del. entire gene | REP1 absent |
| 13216 | | c.1414-1_1414delGAinsAT | splice site | Exon 11 | p.? |
| 13220 | P20 | NA | NA | NA | NA |
| 13224 | | c.757C>T | nonsense | exon 6 | p.Arg253* |
| 13226 | | c.940-2A>T | splice site | Intron 7 | p.? |
| CHM01 | P10 | c.799C>T | nonsense | exon 6 | p.Arg.267* |
| CHM02 | P18 | c.799C>T | nonsense | exon 6 | p.Arg.267* |
| CHM03 | | del. entire gene[††] | deletion | del. entire gene | REP1 absent |
| CHM04 | | c.25_28del4ins11 | frameshift | exon 1 | p.? |
| CHM05 | P11 | NA | NA | NA | NA |
| CHM06 | P12 | c.1138 C>T | nonsense | Exon 9 | p.Gln380* |
| CHM07 | P1 | c.799C>T | nonsense | Exon 6 | p.Arg267* |
| CHM08 | | NA | NA | NA | NA |
| CHM09 | | c.799C>T | nonsense | Exon 6 | p.Arg267* |

NA: sample or screening result not available; C: confirmed mutation positive, resulting mutation not available; del=deletion; p.?: unknown protein effect.

** Novel mutation; [††] Deletion of entire gene and neighboring POU3 gene.

Patients who are siblings or from same pedigree: 13034/13067; 13048/13050; 13057/13183; 13062/13063; 13185/13186;13073/13074;13086/13087;13099/13100;13130/13131/13132;13203/13204/13205;13177/13208; CHM01/CHM02