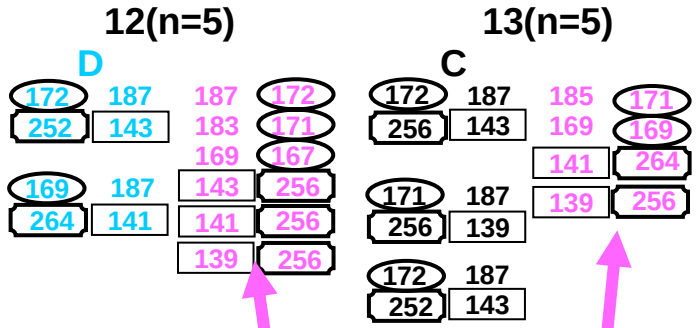
Supporting figure S1 for “Allelic recombination between distinct genomic locations generates copy number diversity in human beta-defensins ” by Suhaili Abu Bakar *et al*.

(28 pages)

Key to Supporting Figure S1 CEPH family data (showing members of family 1350)



Untransmitted grandparental haplotypes shown in **pink**

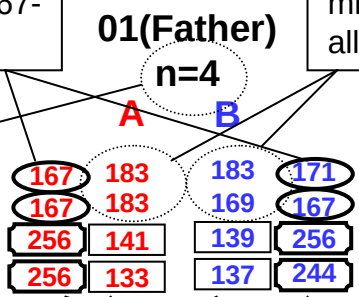
rs5889219 alleles: lengths either in range 119-125 or 167-172

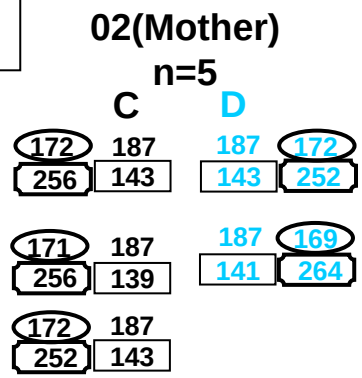
EPEV-1 microsatellite alleles

"n=*" beta-defensin copy number

EPEV-3 microsatellite alleles

EPEV-2 microsatellite alleles (only shown in families 1350 and 1362)

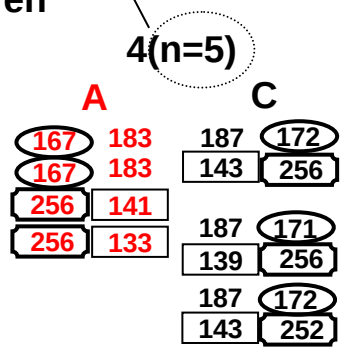


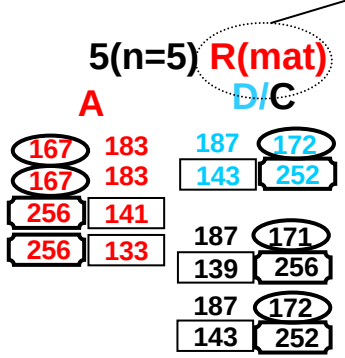


Parental haplotypes: allele combinations in A and B not known, but ordered by crossover in C and D

Recombination of maternal origin: order "D/C" indicates telomeric D/ centromeric C

Children





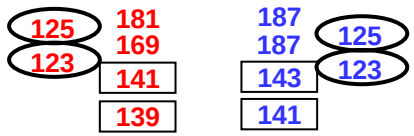
(individual 134612): rs5889219 allele inferred from other loci but not amplified under conditions used

(individual 140813): EPEV-3 allele inferred from other loci but not amplified under conditions used

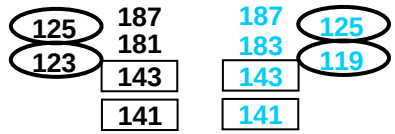
119 (family 35): 119bp rs5889219 product showing consistently reduced PCR yield (50-70% of expected) under conditions used

CEPH
Family 2

01(Father)
n=4

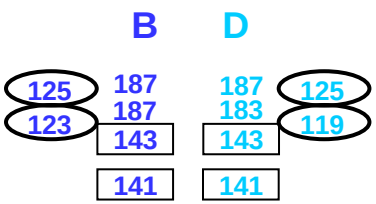


02(Mother)
n=4

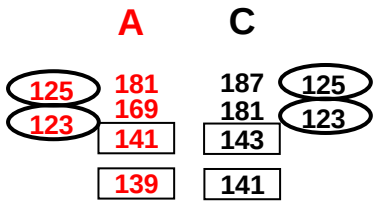


Children

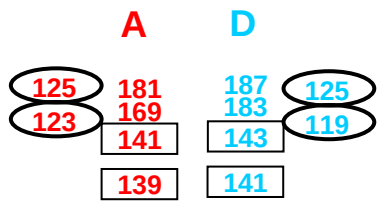
3(n=4)



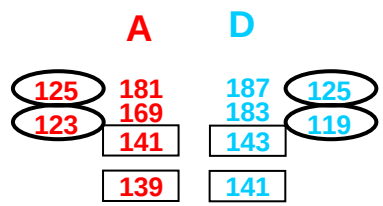
4(n=4)



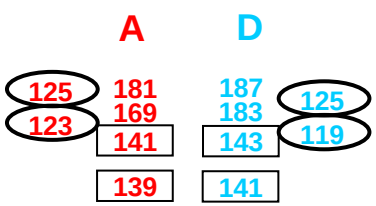
5(n=4)



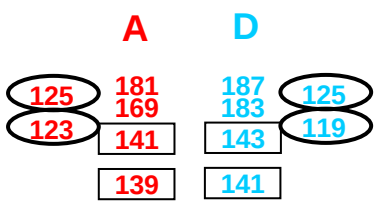
6(n=4)



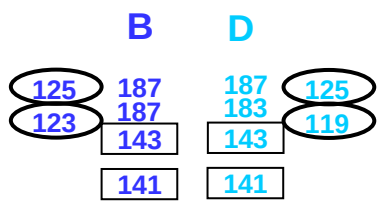
7(n=4)

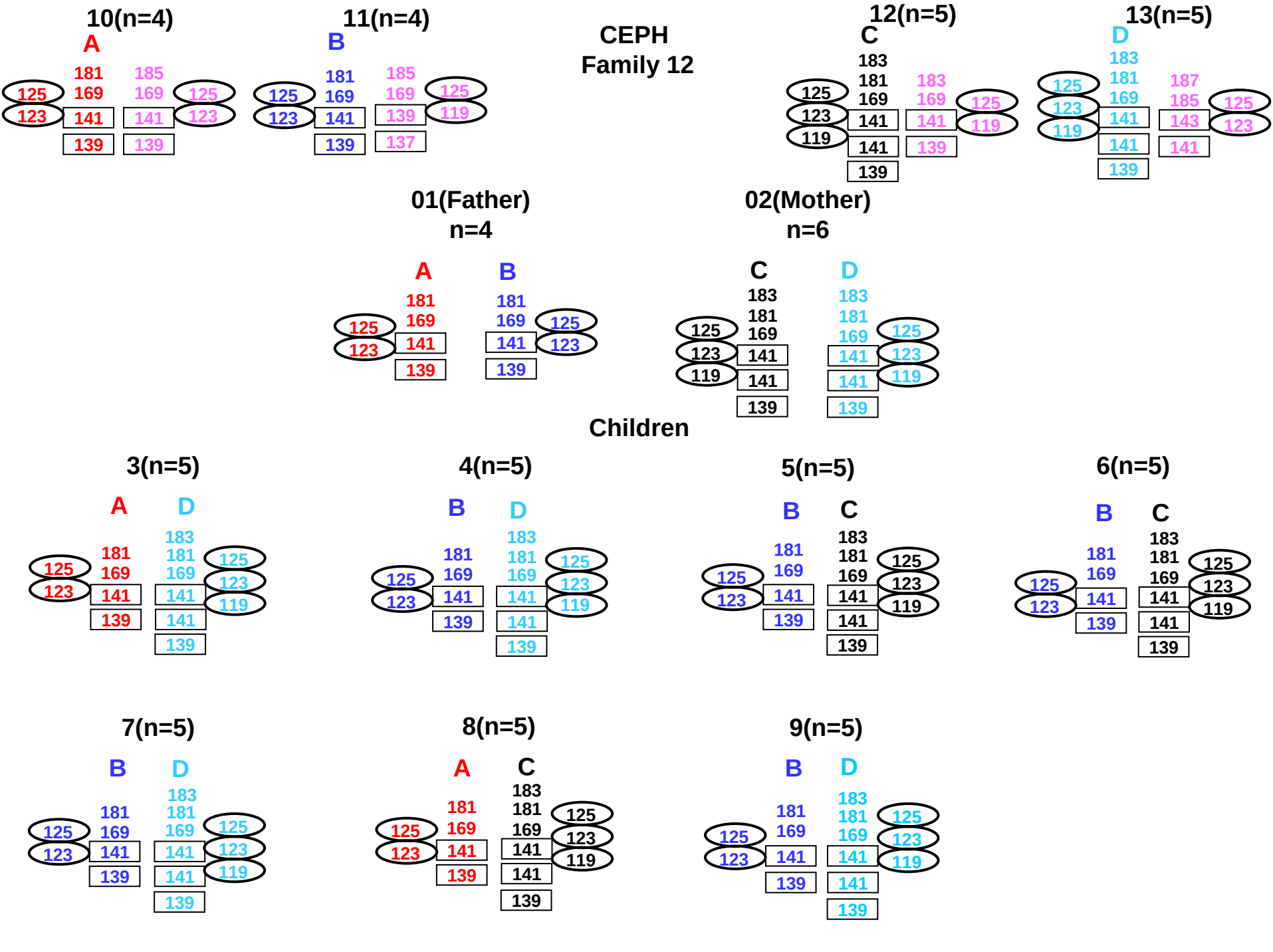


8(n=4)



9(n=4)




10(n=4)
A
181
169
125
123
141
139
185
169
125
141
123
139
11(n=4)
B
181
169
125
123
141
139
185
169
125
139
119
137
CEPH
Family 12
12(n=5)
C
183
181
169
125
123
119
141
141
139
183
169
125
141
119
139
13(n=5)
D
183
181
169
125
123
119
141
141
139
187
185
125
143
123
141
01(Father)
n=4
A
181
169
125
123
141
139
B
181
169
125
141
123
139
02(Mother)
n=6
C
183
181
169
125
123
119
141
141
139
D
183
181
169
125
141
123
141
119
139
Children
3(n=5)
A
181
169
125
123
141
139
D
183
181
169
125
141
123
141
119
139
4(n=5)
B
181
169
125
123
141
139
D
183
181
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125
141
123
141
119
139
5(n=5)
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123
141
139
C
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C
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8(n=5)
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125
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C
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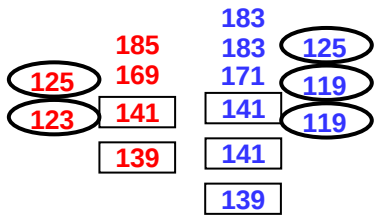
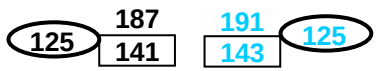
181
169
125
141
123
141
119
139
9(n=5)
B
181
169
125
123
141
139
D
183
181
169
125
141
123
141
119
139

CEPH
Family 23

01(Father)
n=5

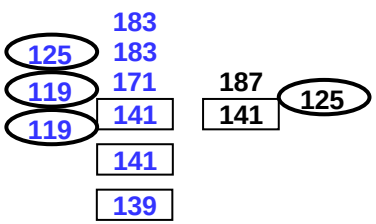


02(Mother)
n=2

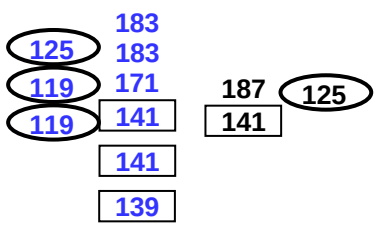


Children

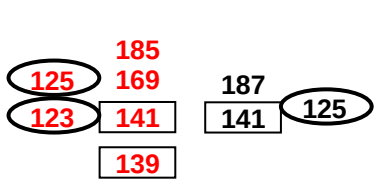
3(n=4)



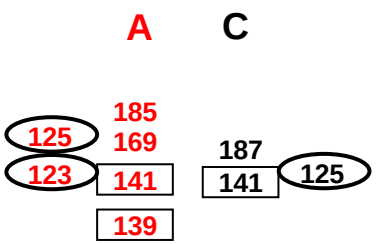
4(n=4)



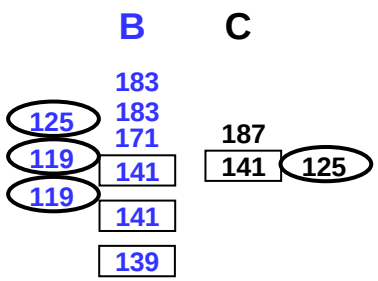
5(n=3)



6(n=3)



7(n=4)



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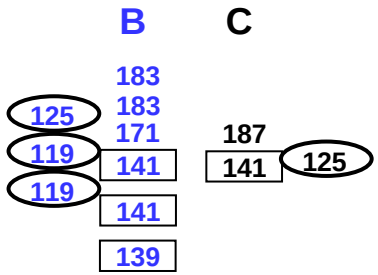
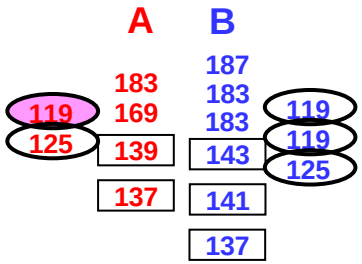
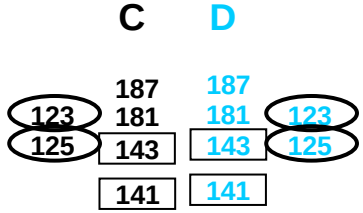


CEPH
Family 35

01(Father)
n=5

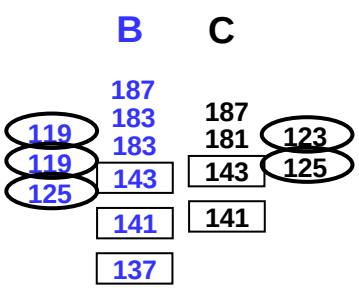


01(Mother)
n=4

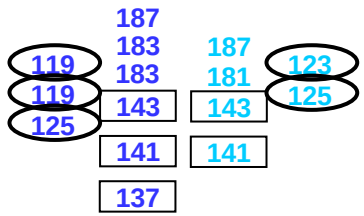


Children

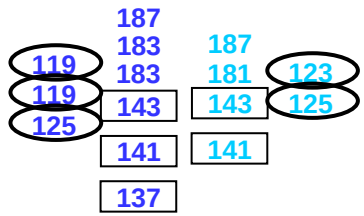
3(n=5)



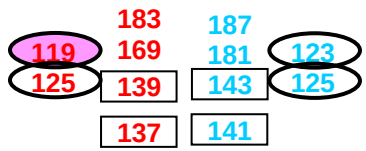
4(n=5)



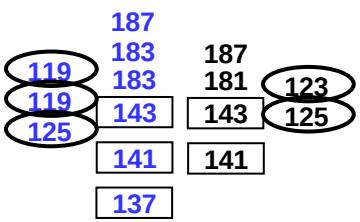
5(n=5)



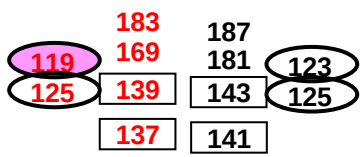
6(n=4)



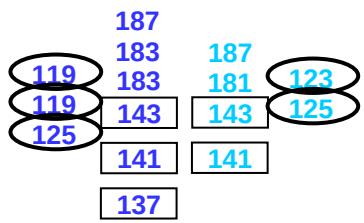
7(n=5)



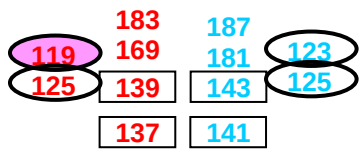
8(n=4)



9(n=5)

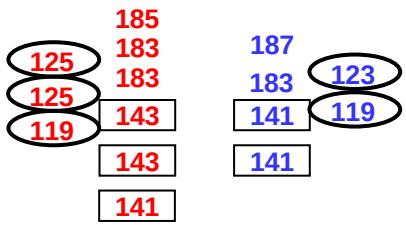


10(n=4)

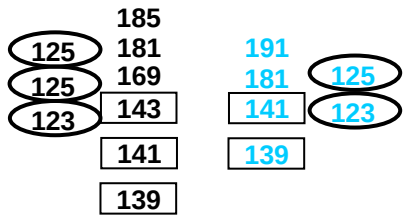


CEPH
Family 37

01(Father)
n=5

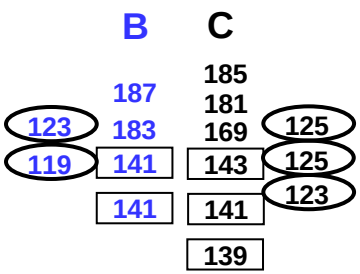


02(Mother)
n=5

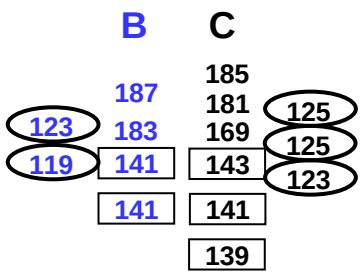


Children

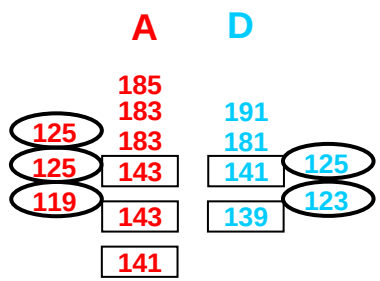
4(n=5)



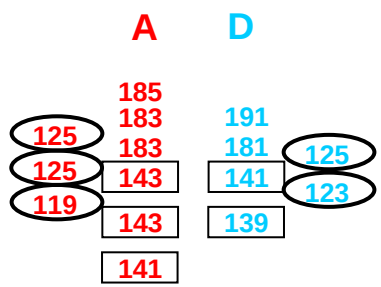
5(n=5)



6(n=5)

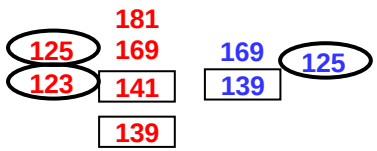


8(n=5)

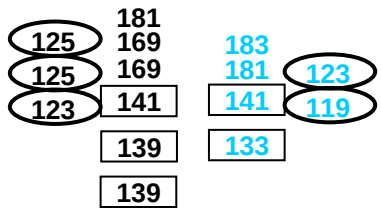


CEPH
Family 45

01(Father)
n=3

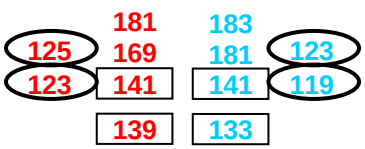


02(Mother)
n=5

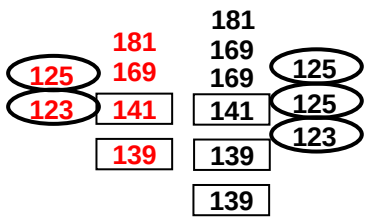


Children

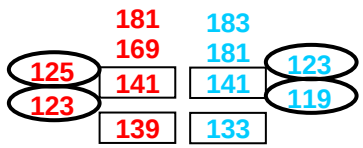
3(n=4)



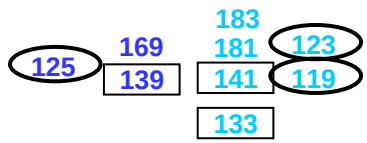
4(n=5)



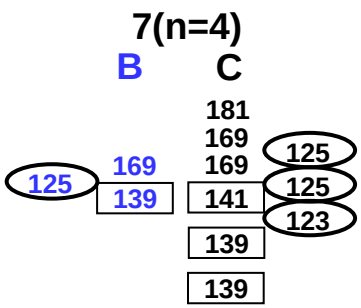
5(n=4)



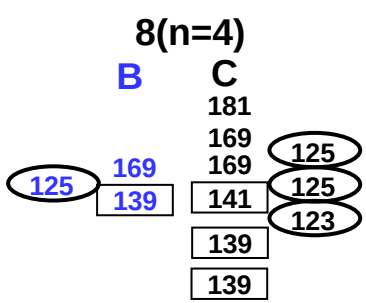
6(n=3)



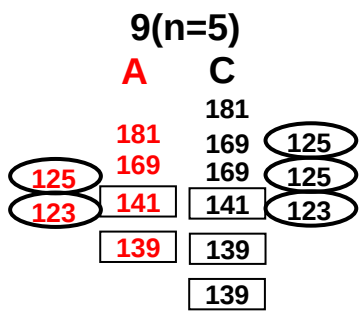
7(n=4)



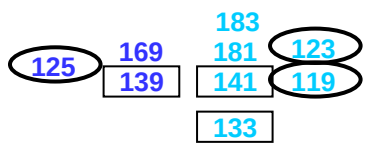
8(n=4)



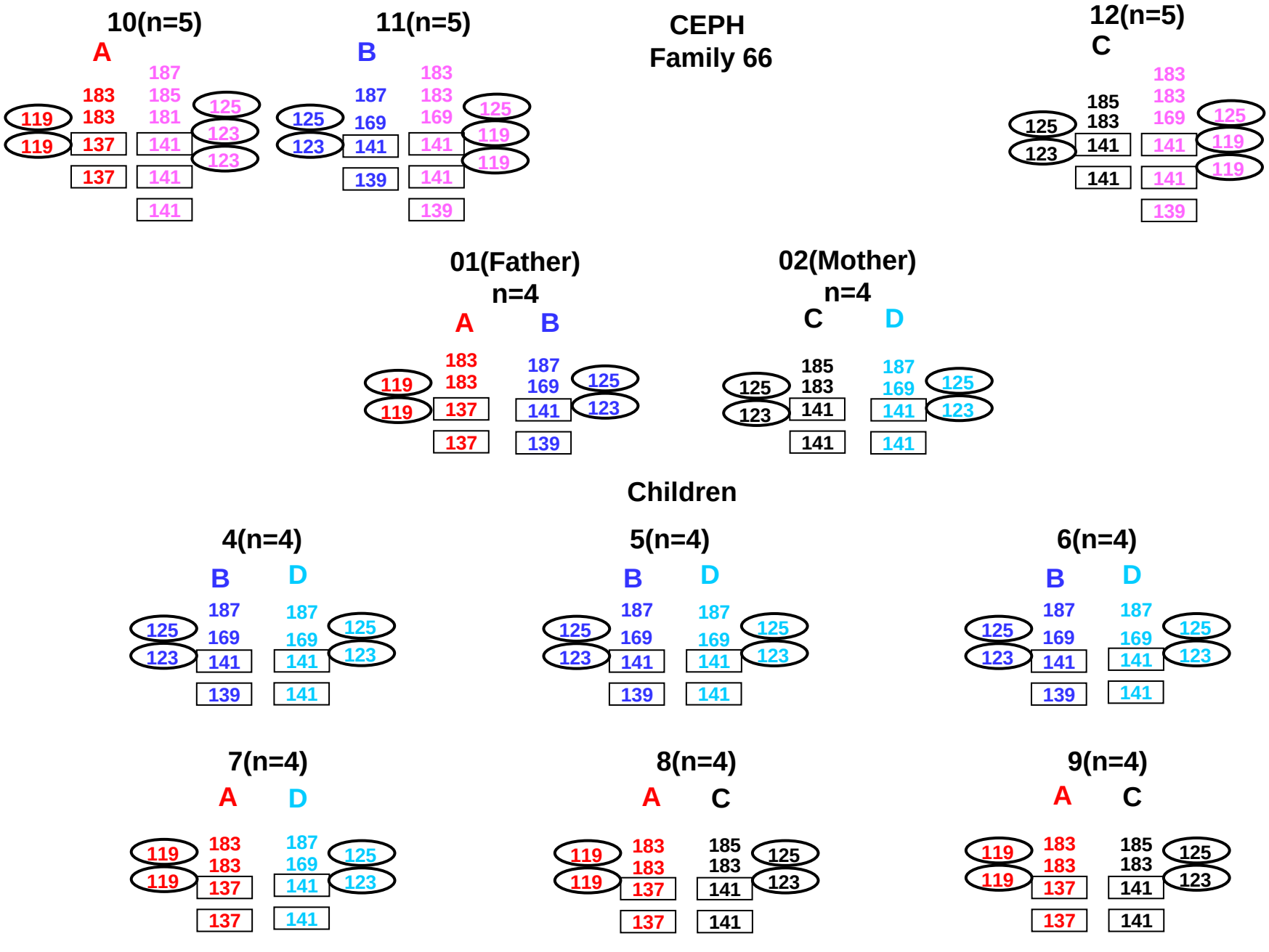
9(n=5)

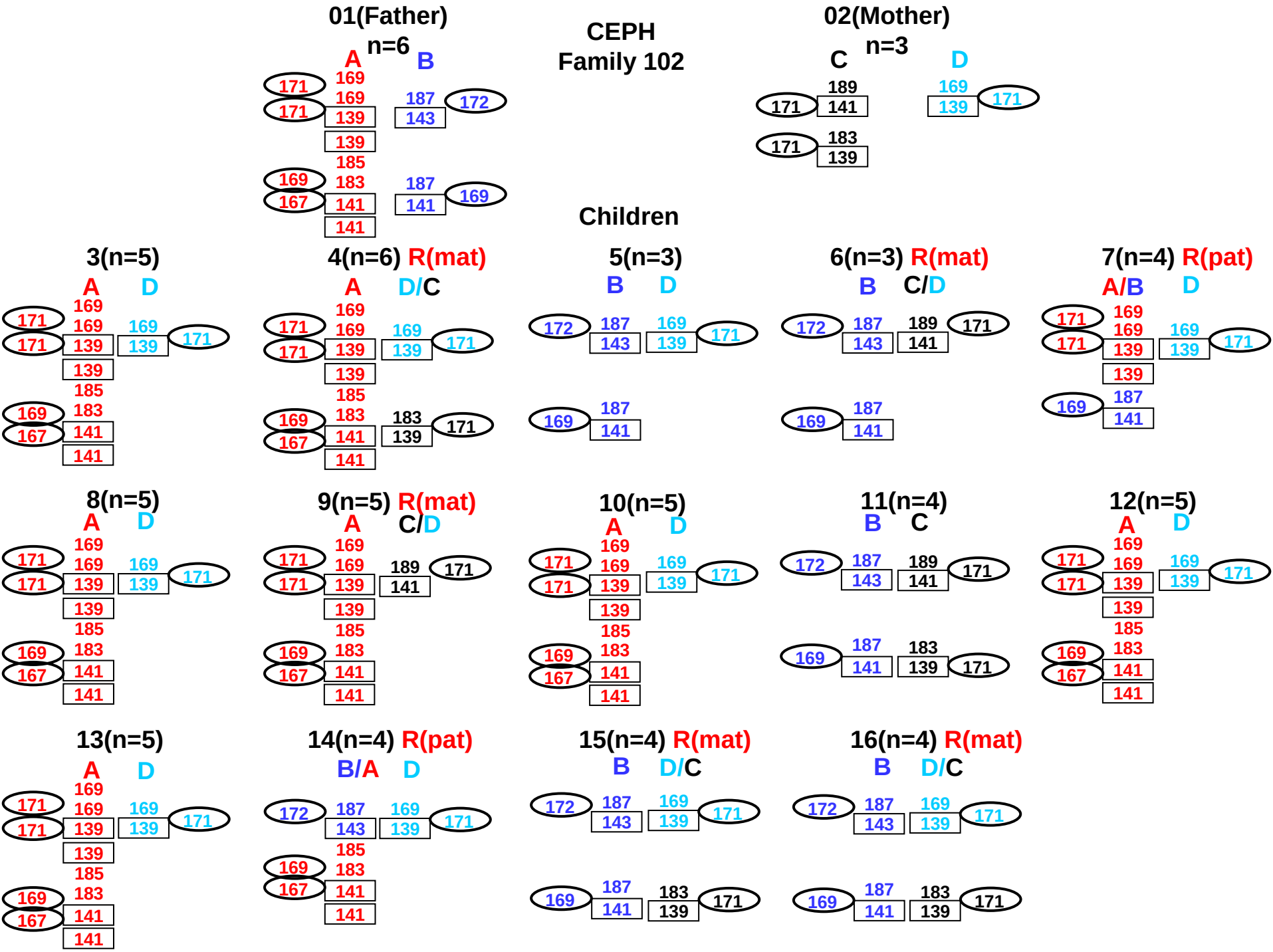


10(n=3)



CEPH Family 66




01(Father)
n=6
A
B
171
169
171
169
139
187
172
139
143
139
185
169
183
187
167
141
141
169
141
CEPH
Family 102
02(Mother)
n=3
C
D
189
171
141
169
171
139
171
183
171
139
Children
3(n=5)
A
D
171
169
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139
139
139
185
169
183
167
141
141
4(n=6) R(mat)
A
D/C
171
169
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169
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183
183
167
141
139
171
141
5(n=3)
B
D
172
187
169
171
143
139
169
187
141
6(n=3) R(mat)
B
C/D
172
187
189
171
143
141
169
187
141
7(n=4) R(pat)
A/B
D
171
169
169
171
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139
171
139
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187
141
8(n=5)
A
D
171
169
171
169
139
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183
167
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141
9(n=5) R(mat)
A
C/D
171
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171
189
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139
141
139
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183
167
141
141
10(n=5)
A
D
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171
169
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169
171
139
139
139
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169
183
167
141
141
11(n=4)
B
C
172
187
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171
143
141
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12(n=5)
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D
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13(n=5)
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D
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139
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169
183
167
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14(n=4) R(pat)
B/A
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183
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15(n=4) R(mat)
B
D/C
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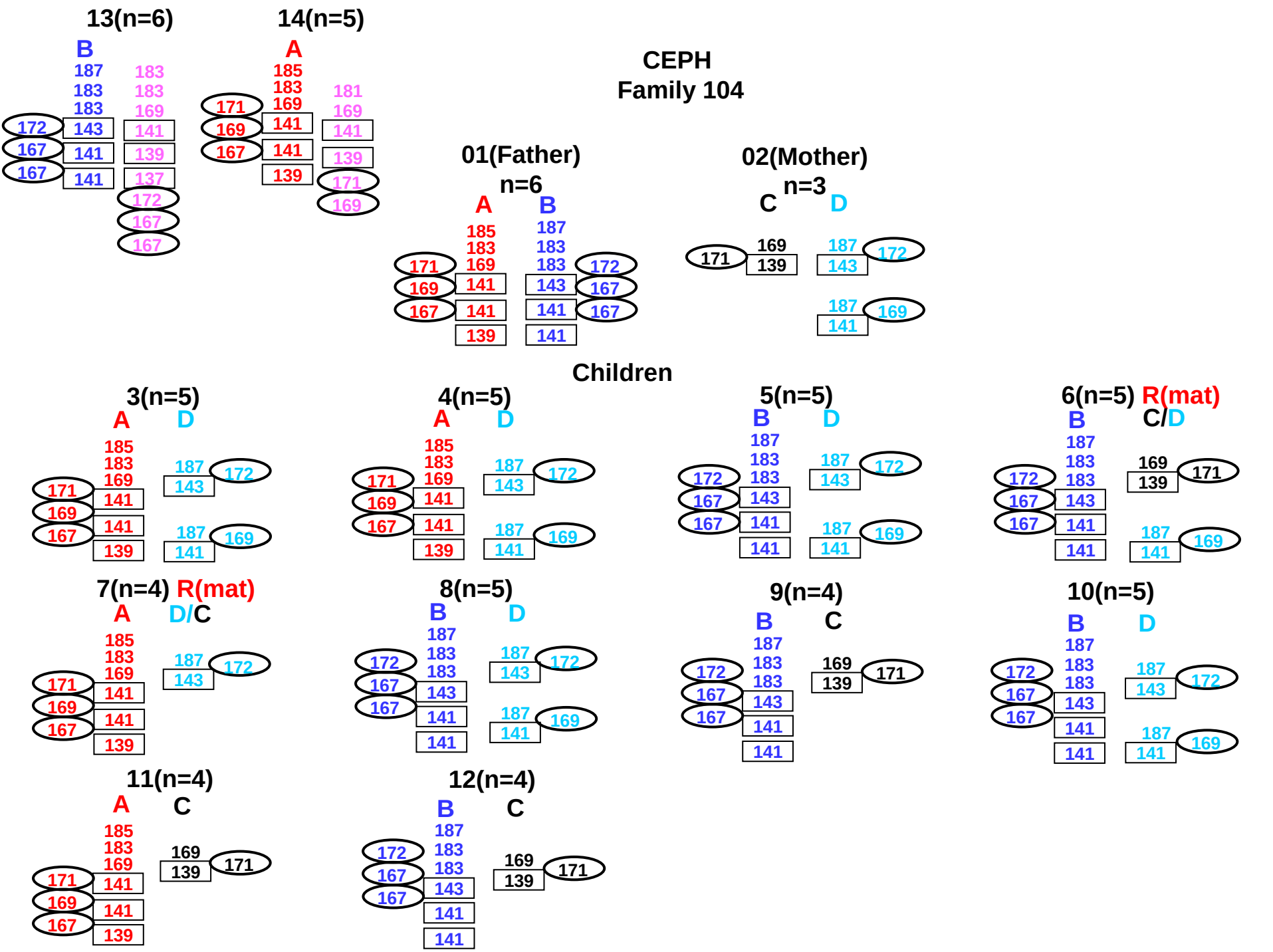
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14(n=5)
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185
183
169
171
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171
169
CEPH
Family 104
01(Father)
n=6
A
B
185
183
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169
167
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141
02(Mother)
n=3
C
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172
187
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169
Children
3(n=5)
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4(n=5)
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172
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169
5(n=5)
B
D
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183
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167
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143
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141
169
6(n=5) R(mat)
B
C/D
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183
183
172
167
167
143
141
141
169
139
171
187
141
169
7(n=4) R(mat)
A
D/C
185
183
169
171
169
167
141
141
139
187
143
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8(n=5)
B
D
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141
187
143
172
187
141
169
9(n=4)
B
C
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183
183
172
167
167
143
141
141
169
139
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10(n=5)
B
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169
11(n=4)
A
C
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139
169
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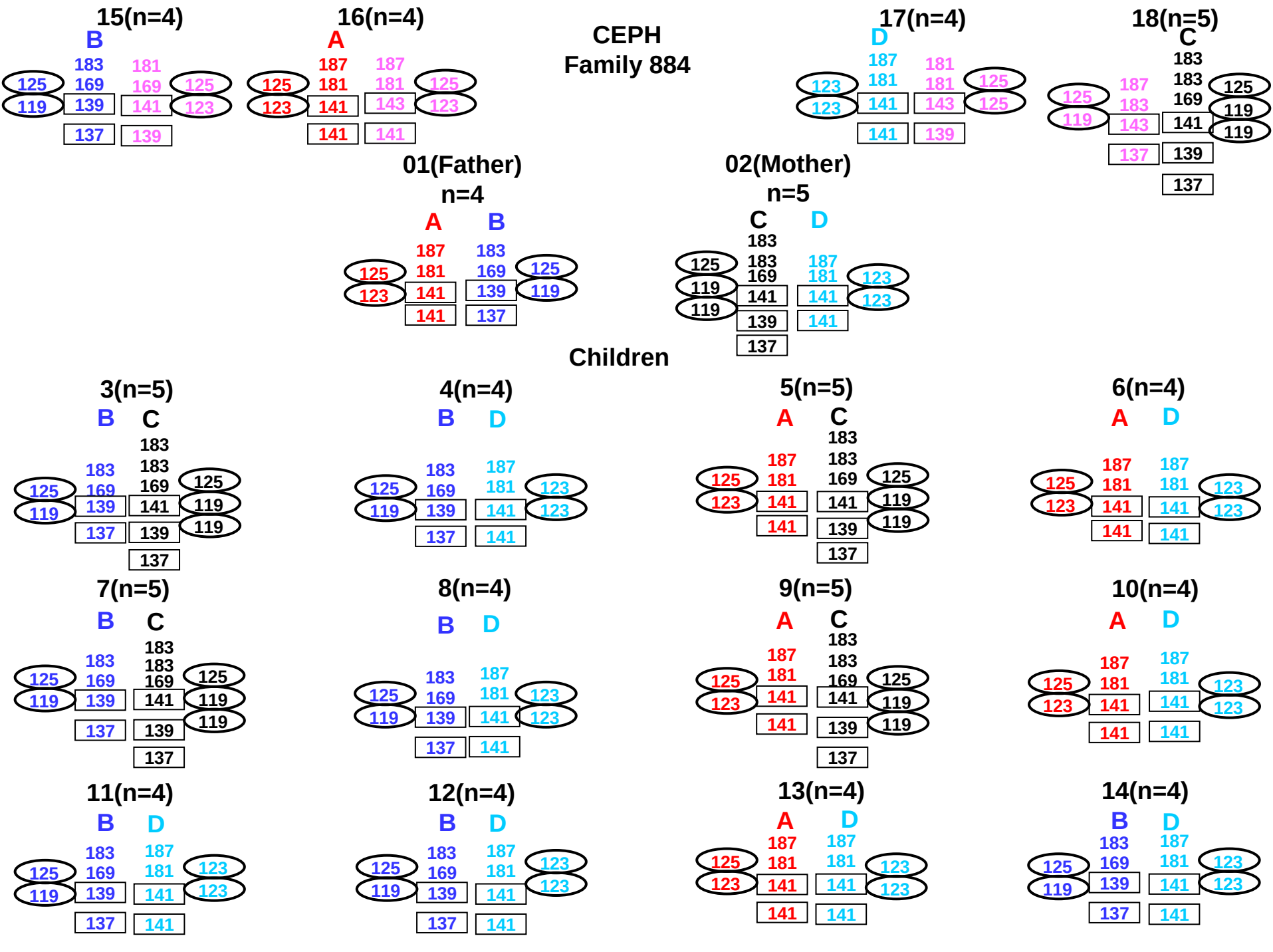
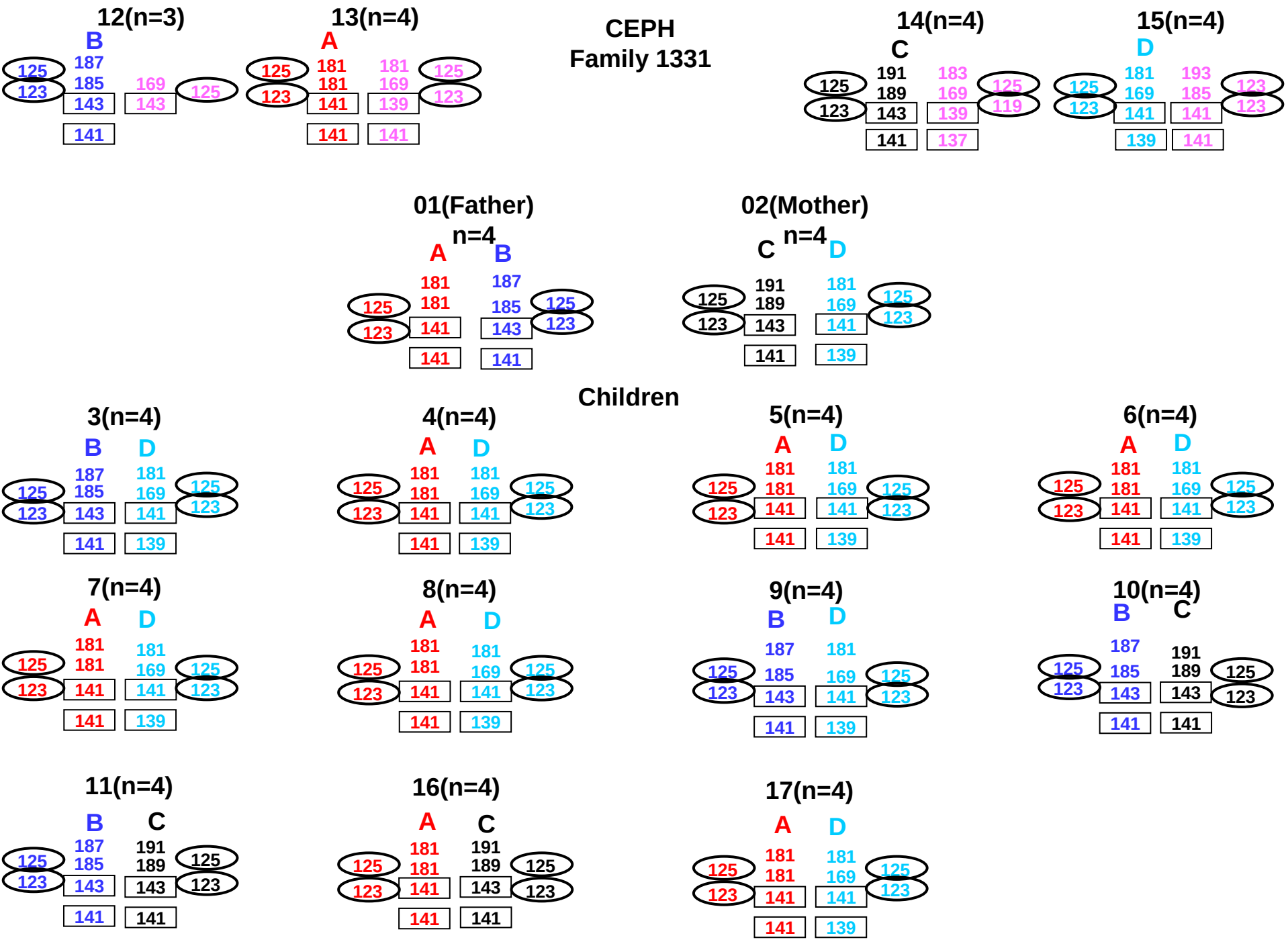
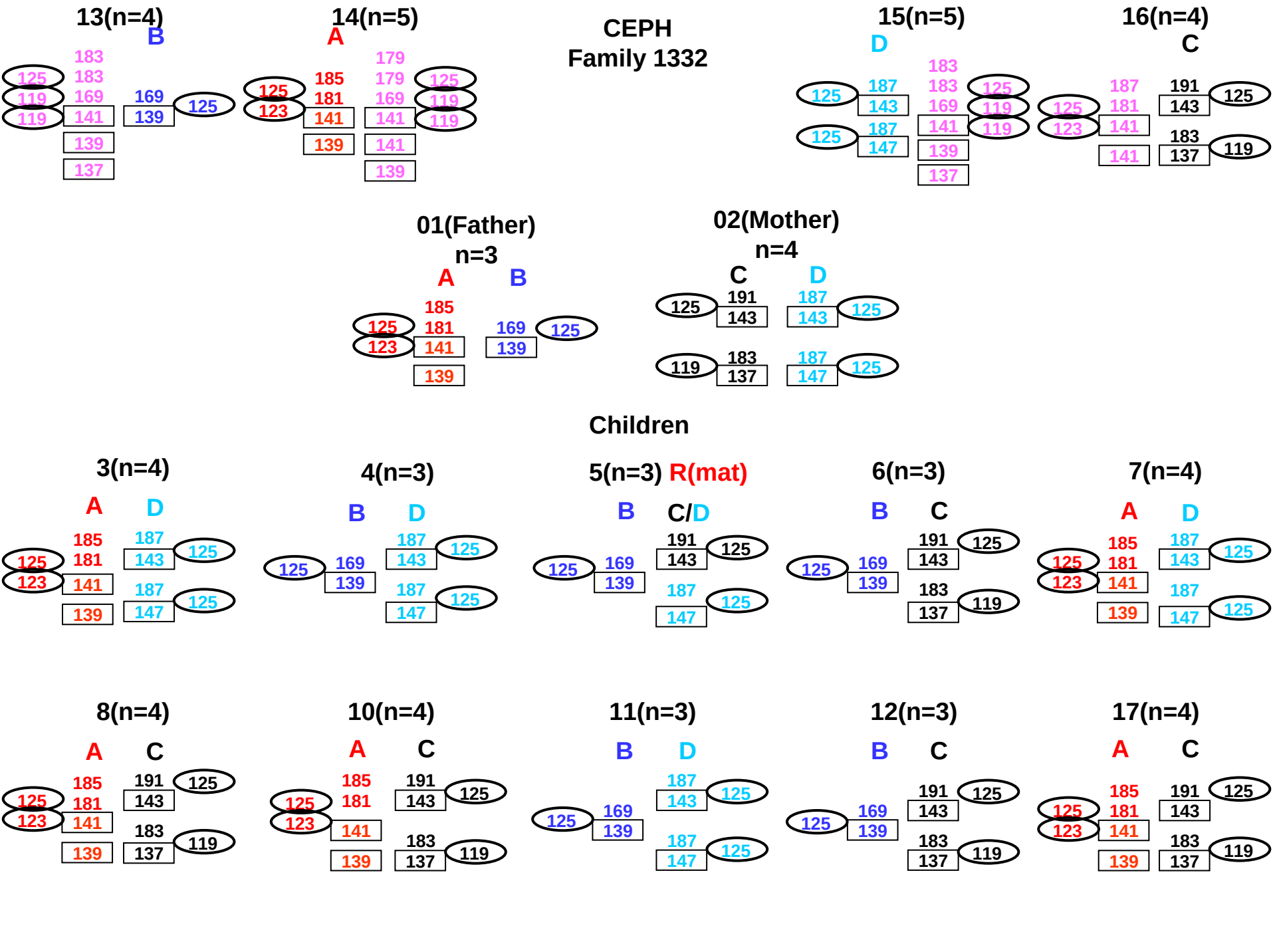
171

CEPH Family 884



CEPH Family 1331

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14(n=5)
A
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169
141
141
139
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119
185
181
141
139
125
123
CEPH
Family 1332
15(n=5)
D
183
183
169
141
139
137
125
119
119
187
143
187
147
125
125
16(n=4)
C
191
143
183
137
187
181
141
141
125
123
125
119
01(Father)
n=3
A
B
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139
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02(Mother)
n=4
C
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A
D
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141
139
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123
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143
187
147
125
125
4(n=3)
B
D
169
139
187
143
187
147
125
125
5(n=3) R(mat)
B
C/D
169
139
191
143
187
147
125
125
6(n=3)
B
C
169
139
191
143
183
137
125
125
119
7(n=4)
A
D
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147
125
125
123
125
8(n=4)
A
C
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183
137
125
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123
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10(n=4)
A
C
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139
191
143
183
137
125
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123
119
11(n=3)
B
D
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139
187
143
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125
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12(n=3)
B
C
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191
143
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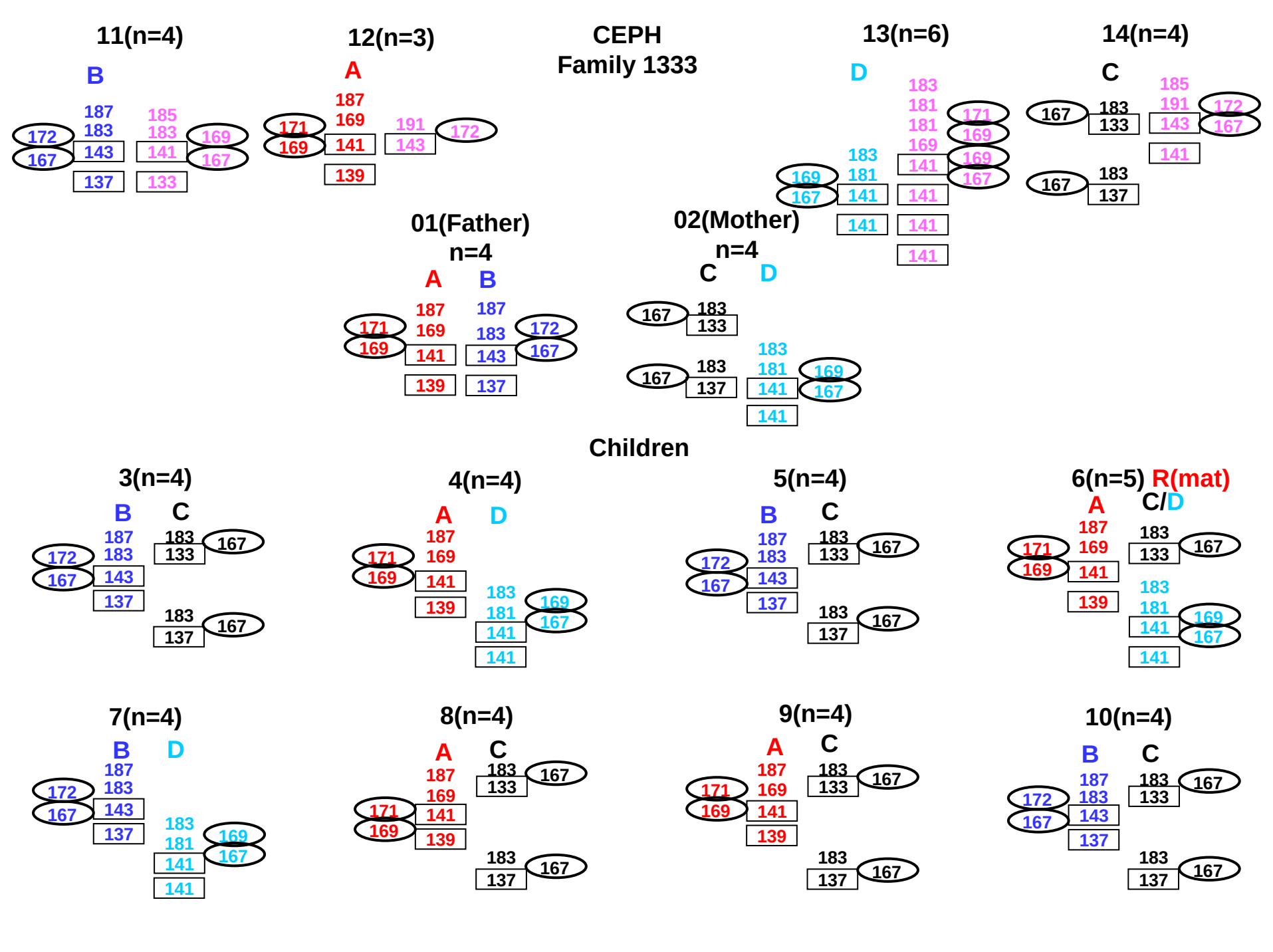
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B
187
183
172
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137
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183
141
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167
133
12(n=3)
A
187
169
171
141
169
139
191
143
172
CEPH
Family 1333
13(n=6)
D
183
181
171
181
169
169
141
183
169
181
169
141
167
141
141
141
14(n=4)
C
167
183
133
185
191
172
143
167
141
167
183
137
01(Father)
n=4
A
B
187
187
171
169
183
172
169
141
143
167
139
137
02(Mother)
n=4
C
D
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133
183
181
169
167
183
137
141
167
141
Children
3(n=4)
B
C
187
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137
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133
167
183
137
167
4(n=4)
A
D
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169
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183
181
169
167
141
141
5(n=4)
B
C
187
183
172
143
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137
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133
167
183
137
167
6(n=5) R(mat)
A
C/D
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171
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169
141
139
183
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7(n=4)
B
D
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137
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8(n=4)
A
C
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183
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169
139
183
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167
9(n=4)
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133
167
169
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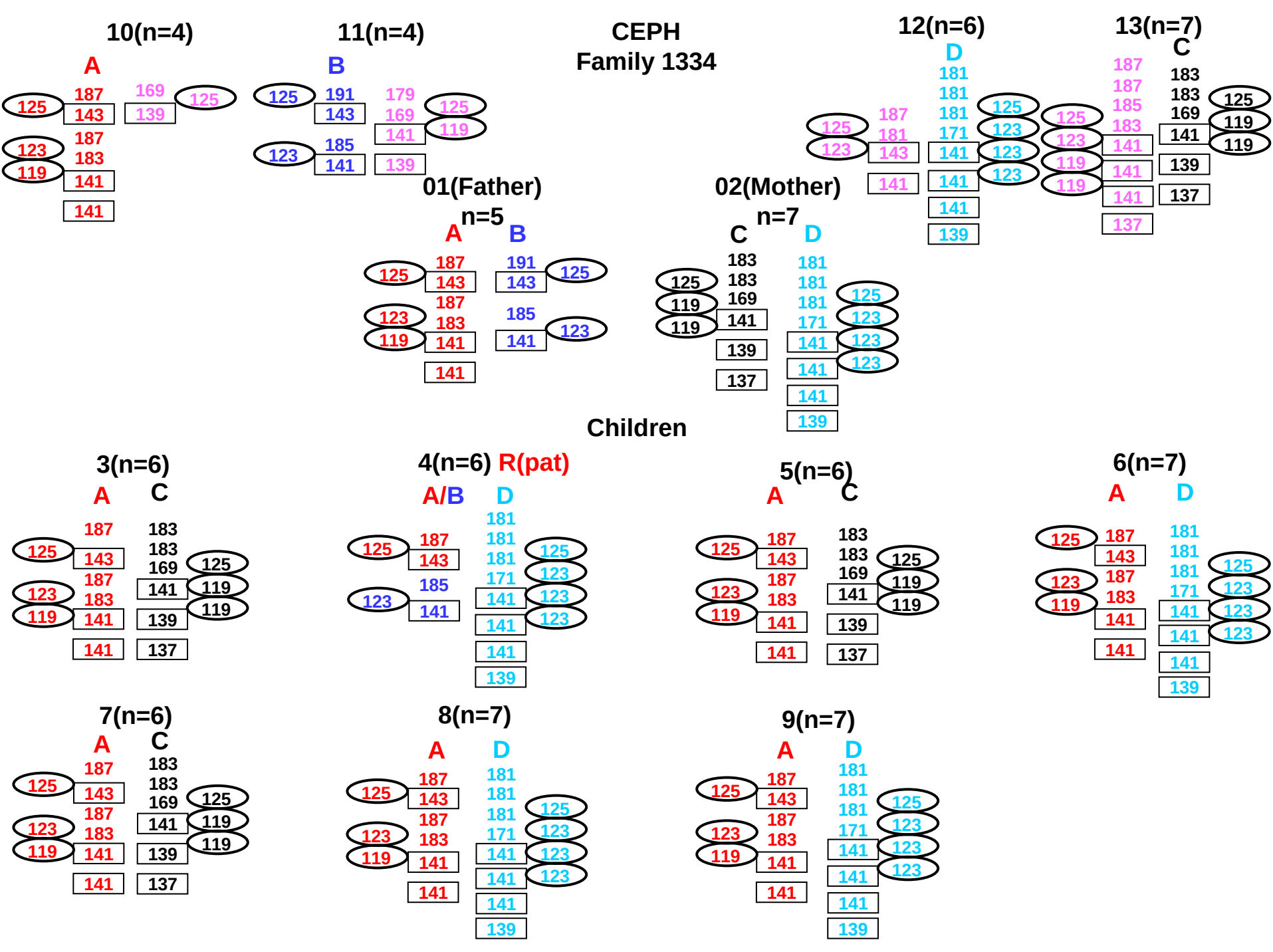
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123
119
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11(n=4)
B
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179
169
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123
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141
139
12(n=6)
D
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187
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13(n=7)
C
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187
185
183
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169
141
139
137
125
119
119
01(Father)
n=5
A
B
187
143
191
143
125
125
187
183
141
123
119
141
185
141
123
141
02(Mother)
n=7
C
D
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183
169
141
139
137
181
181
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171
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Children
3(n=6)
A
C
187
143
187
183
141
123
119
141
183
183
169
141
139
137
125
125
119
119
4(n=6) R(pat)
A/B
D
187
143
185
141
123
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181
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139
125
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123
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5(n=6)
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C
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169
141
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6(n=7)
A
D
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C
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8(n=7)
A
D
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123
123
9(n=7)
A
D
187
143
187
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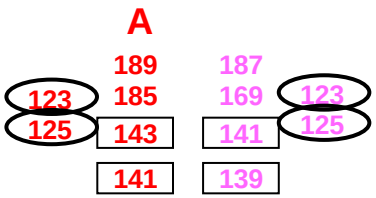
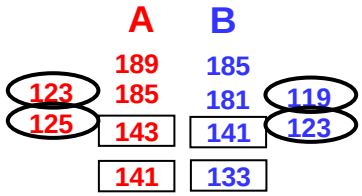
CEPH
Family 1341

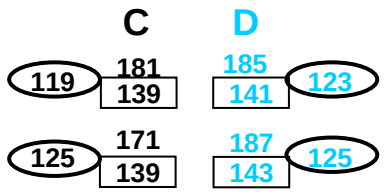
11(n=4)



01(Father) n=4

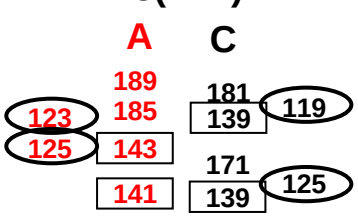


02(Mother) n=4

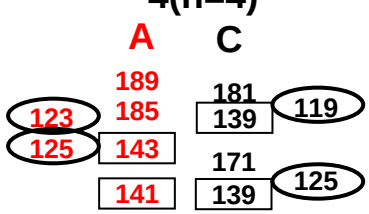


Children

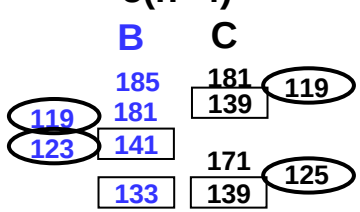
3(n=4)



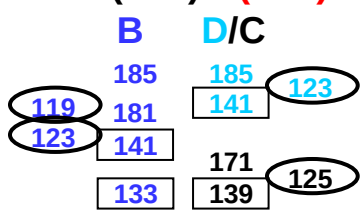
4(n=4)



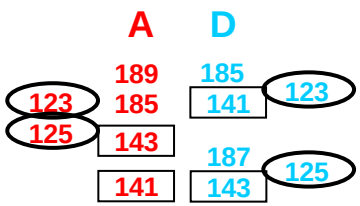
5(n=4)



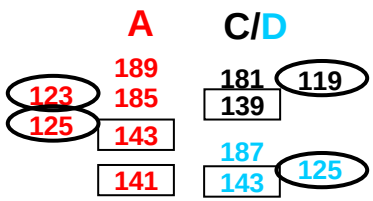
6(n=4) R(mat)



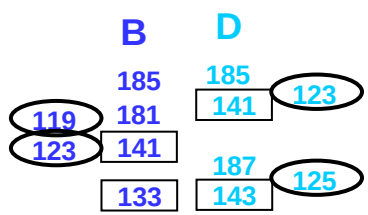
8(n=4)



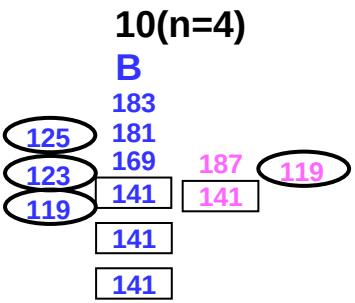
9(n=4) R(mat)

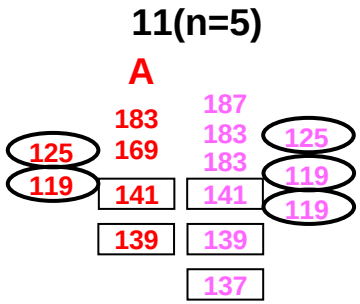


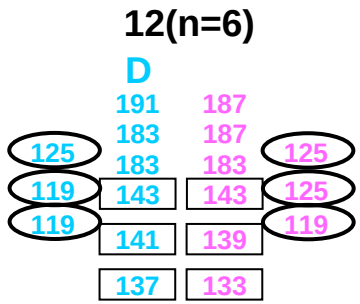
10(n=4)

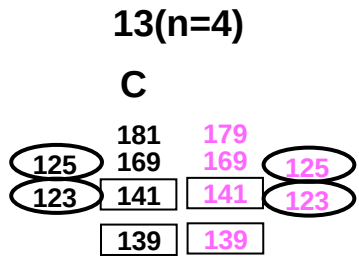


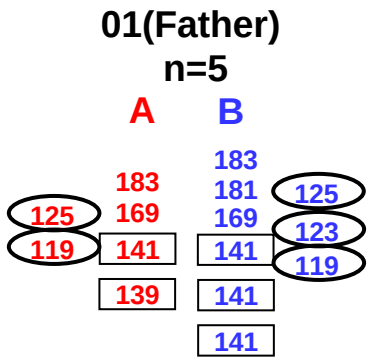
CEPH
Family 1345

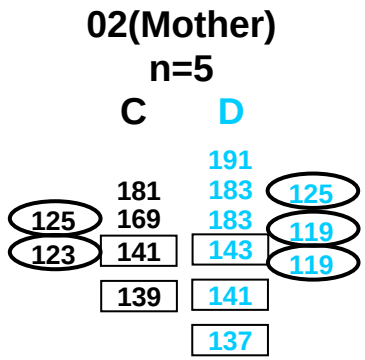




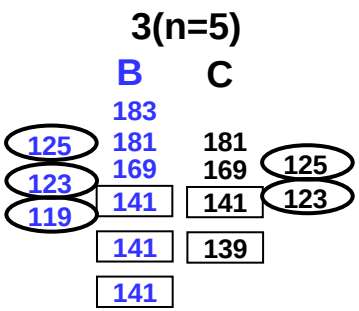


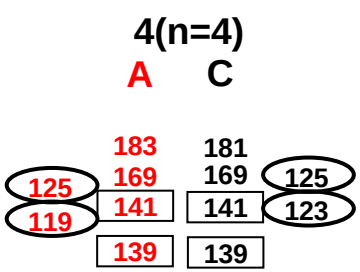


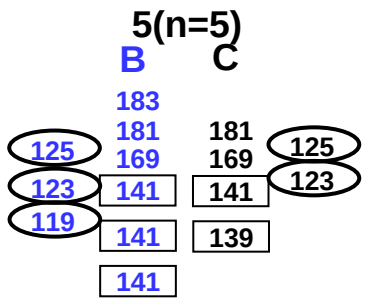


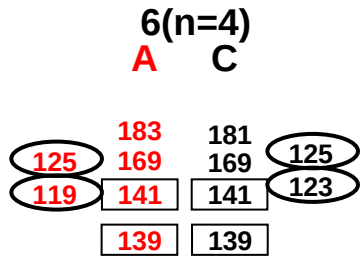


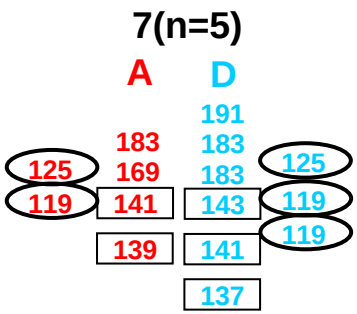
Children

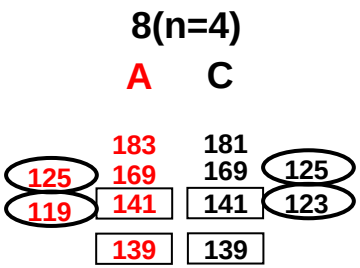


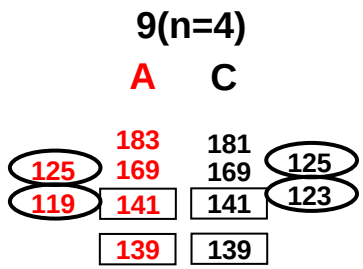




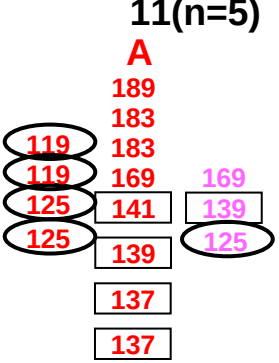


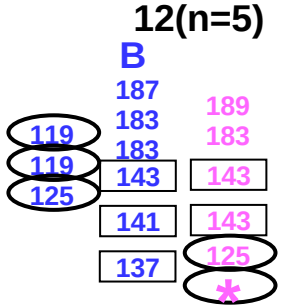


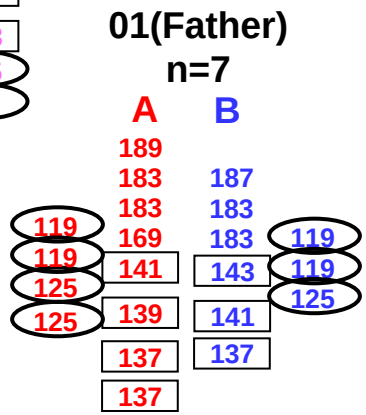


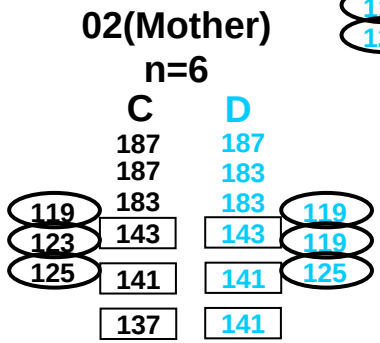


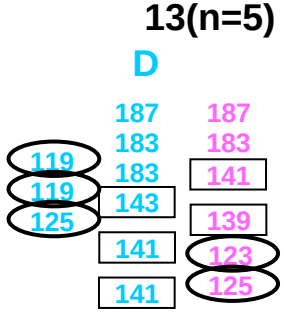
CEPH
Family 1346

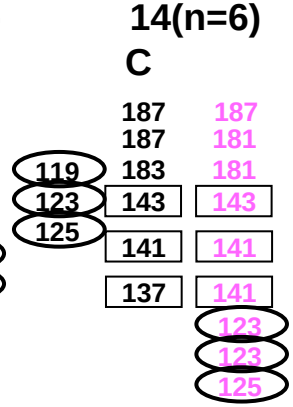




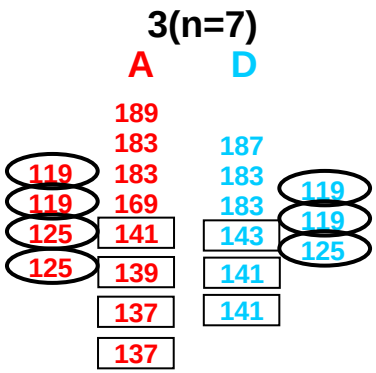


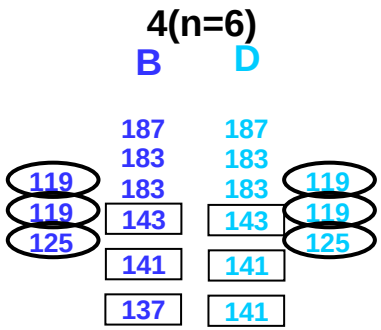


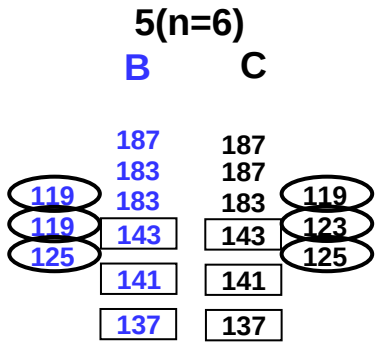


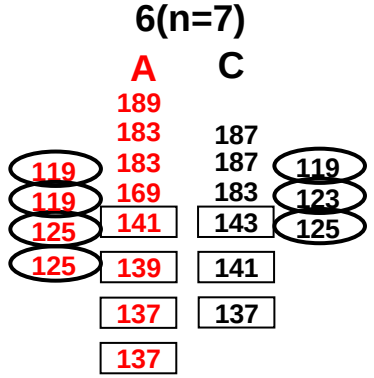


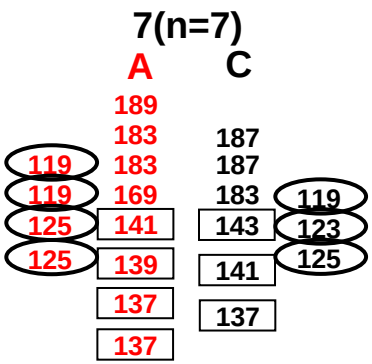
Children

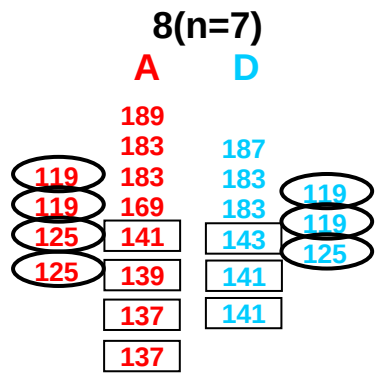


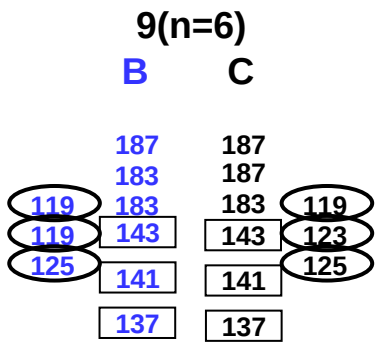


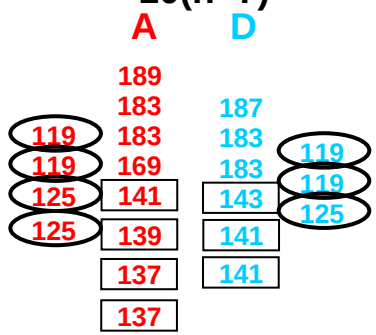


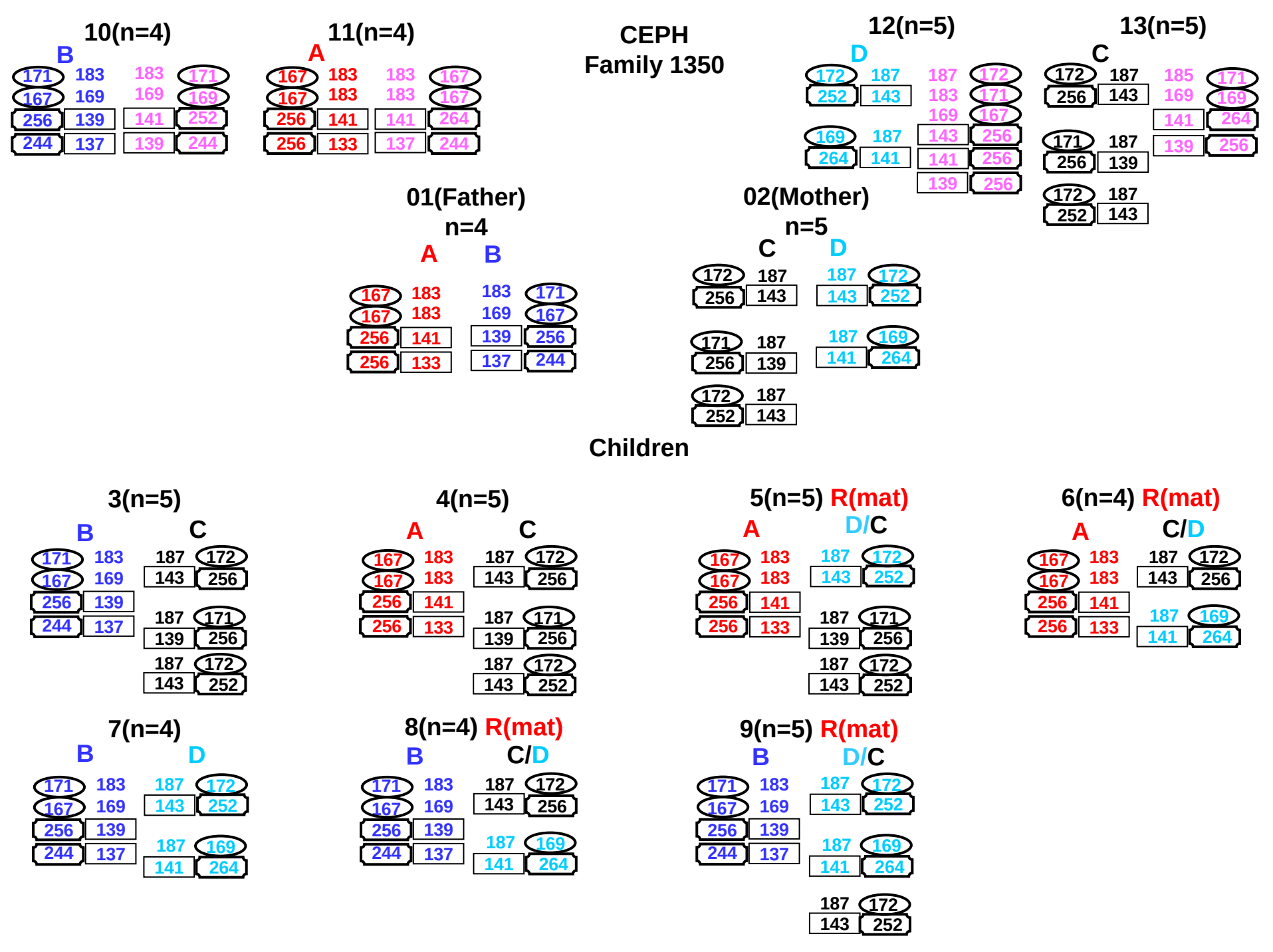


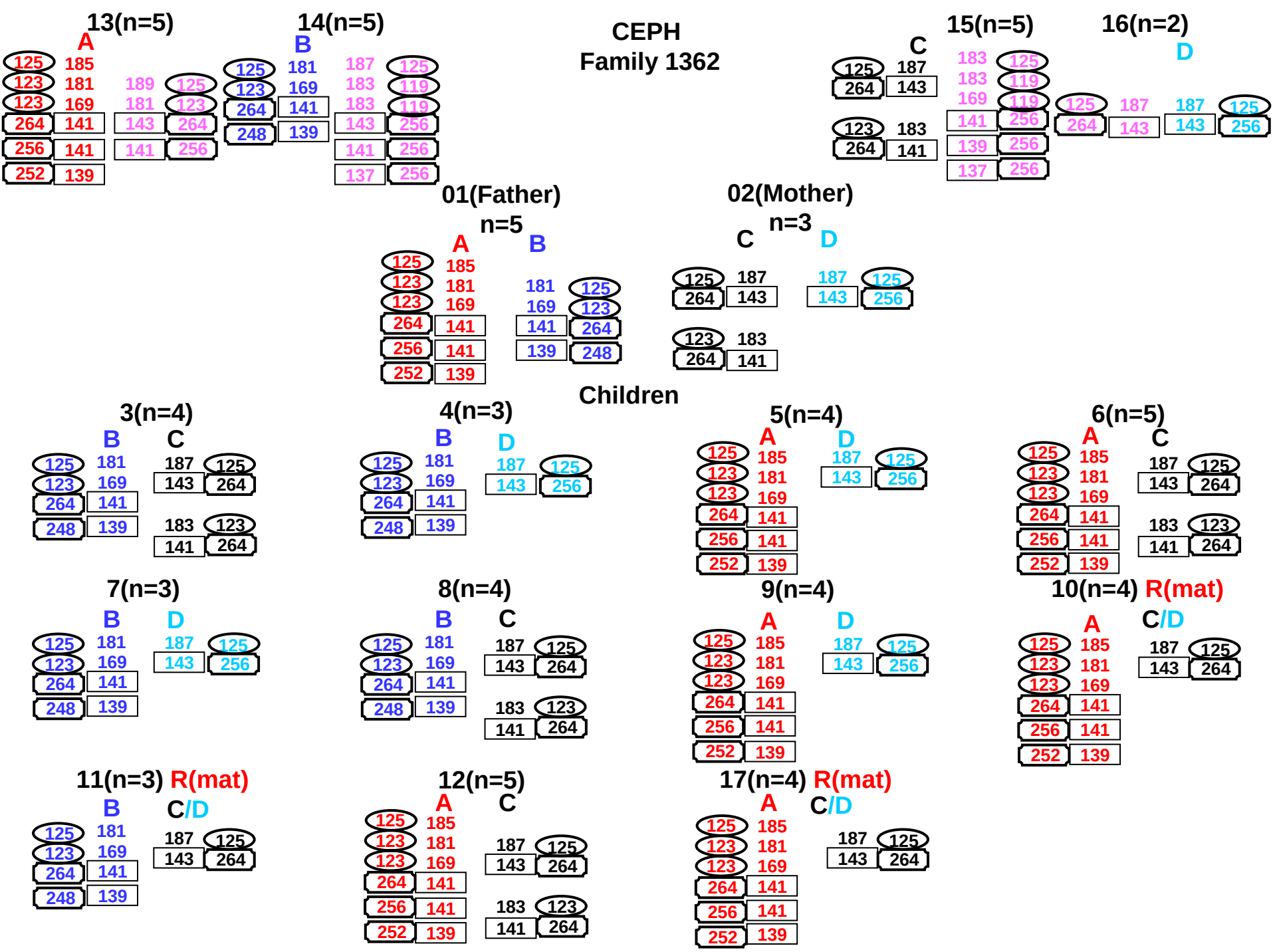




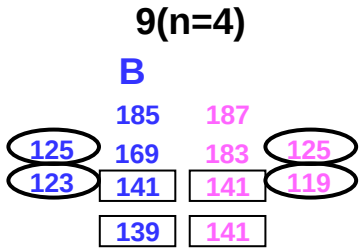


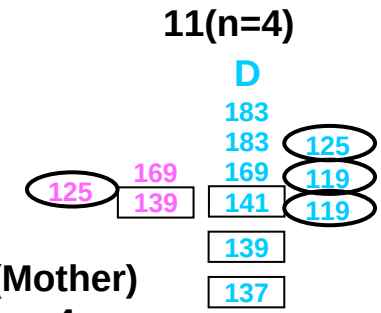


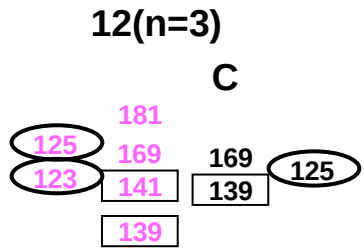

10(n=4)
B
171 183 183 171
167 169 169 169
256 139 141 252
244 137 139 244
11(n=4)
A
167 183 183 167
167 183 183 167
256 141 141 264
256 133 137 244
CEPH
Family 1350
12(n=5)
D
172 187 187 172
252 143 183 171
169 167
169 187 143 256
264 141 141 256
139 256
13(n=5)
C
172 187 185 171
256 143 169 169
141 264
171 187 139 256
256 139
172 187
252 143
01(Father)
n=4
A B
167 183 183 171
167 183 169 167
256 141 139 256
256 133 137 244
02(Mother)
n=5
C D
172 187 187 172
256 143 143 252
171 187 187 169
256 139 141 264
172 187
252 143
Children
3(n=5)
B C
171 183 187 172
167 169 143 256
256 139 187 171
244 137 139 256
187 172
143 252
4(n=5)
A C
167 183 187 172
167 183 143 256
256 141 187 171
256 133 139 256
187 172
143 252
5(n=5) R(mat)
A D/C
167 183 187 172
167 183 143 252
256 141 187 171
256 133 139 256
187 172
143 252
6(n=4) R(mat)
A C/D
167 183 187 172
167 183 143 256
256 141 187 169
256 133 141 264
7(n=4)
B D
171 183 187 172
167 169 143 252
256 139 187 169
244 137 141 264
8(n=4) R(mat)
B C/D
171 183 187 172
167 169 143 256
256 139 187 169
244 137 141 264
9(n=5) R(mat)
B D/C
171 183 187 172
167 169 143 252
256 139 187 169
244 137 141 264
187 172
143 252

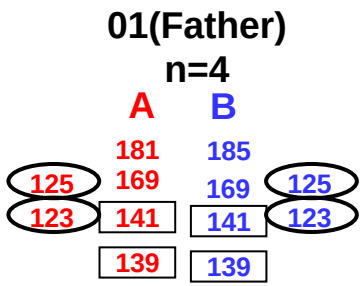

13(n=5)
A
125 185
123 181
123 169
264 141
256 141
252 139
189 125
181 123
143 264
141 256
14(n=5)
B
125 181
123 169
264 141
248 139
187 125
183 119
143 256
141 256
137 256
CEPH
Family 1362
15(n=5)
C
125 187
264 143
123 183
264 141
183 125
183 119
169 119
141 256
139 256
137 256
16(n=2)
D
125 187 187 125
264 143 143 256
01(Father)
n=5
A
125 185
123 181
123 169
264 141
256 141
252 139
B
181 125
169 123
141 264
139 248
02(Mother)
n=3
C
125 187
264 143
123 183
264 141
D
187 125
143 256
Children
3(n=4)
B
125 181
123 169
264 141
248 139
C
187 125
143 264
183 123
141 264
4(n=3)
B
125 181
123 169
264 141
248 139
D
187 125
143 256
5(n=4)
A
125 185
123 181
123 169
264 141
256 141
252 139
D
187 125
143 256
6(n=5)
A
125 185
123 181
123 169
264 141
256 141
252 139
C
187 125
143 264
183 123
141 264
7(n=3)
B
125 181
123 169
264 141
248 139
D
187 125
143 256
8(n=4)
B
125 181
123 169
264 141
248 139
C
187 125
143 264
183 123
141 264
9(n=4)
A
125 185
123 181
123 169
264 141
256 141
252 139
D
187 125
143 256
10(n=4) R(mat)
A
125 185
123 181
123 169
264 141
256 141
252 139
C/D
187 125
143 264
11(n=3) R(mat)
B
125 181
123 169
264 141
248 139
C/D
187 125
143 264
12(n=5)
A
125 185
123 181
123 169
264 141
256 141
252 139
C
187 125
143 264
183 123
141 264
17(n=4) R(mat)
A
125 185
123 181
123 169
264 141
256 141
252 139
C/D
187 125
143 264

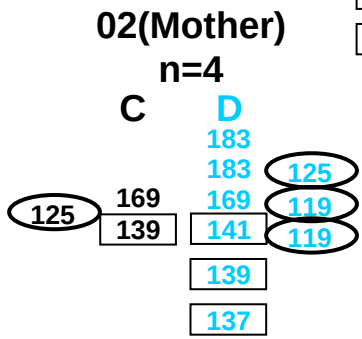
CEPH Family 1375



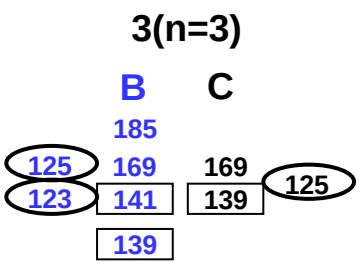


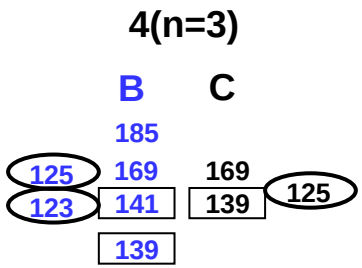


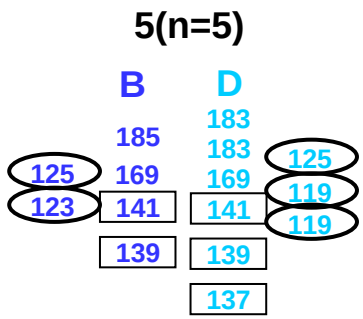


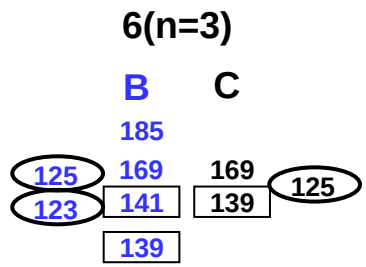


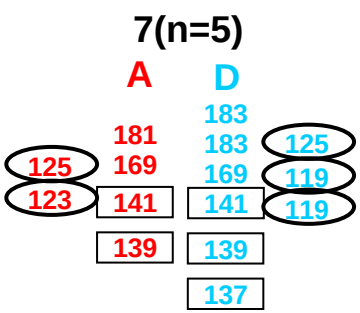
Children

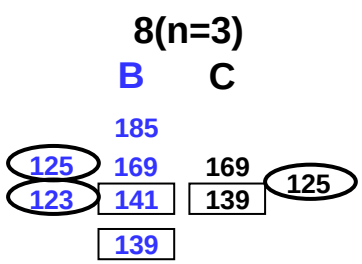


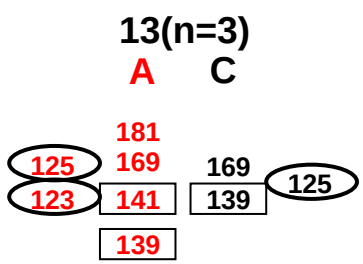




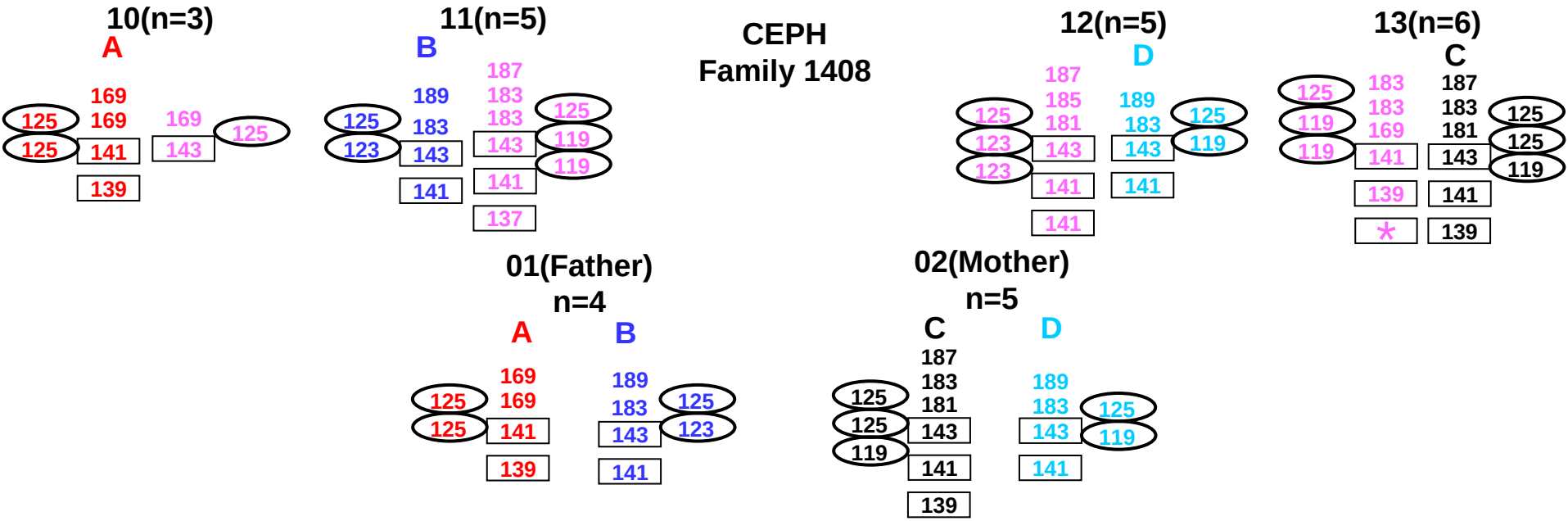




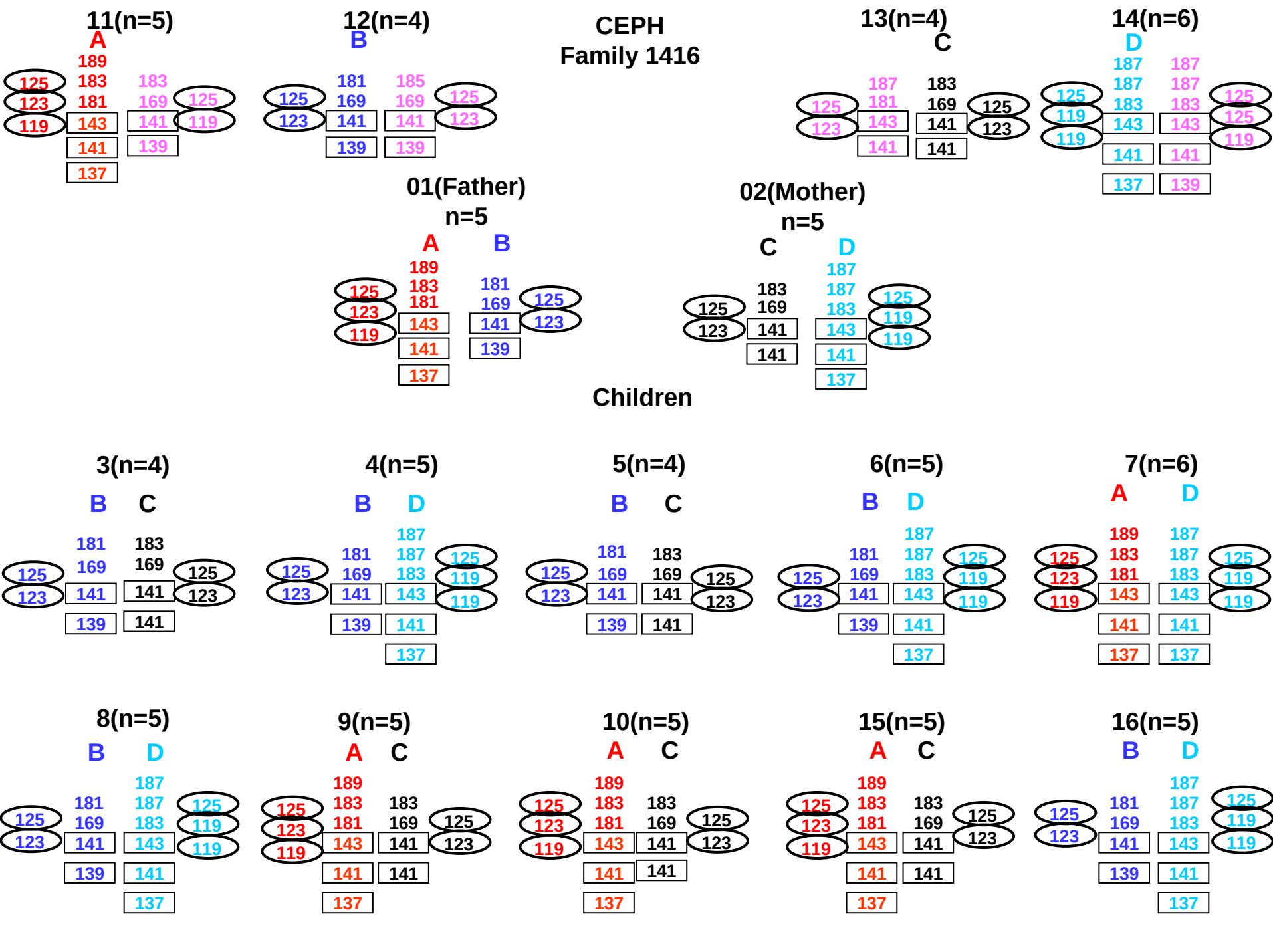




CEPH Family 1408

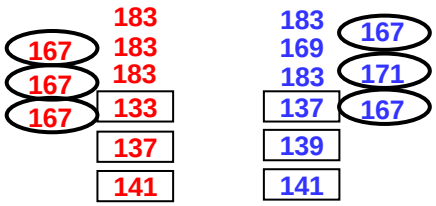


CEPH Family 1416

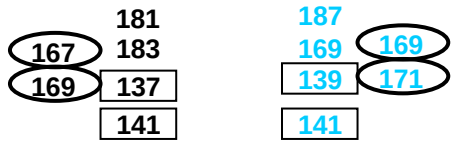


CEPH
Family 1421

01(Father)
n=6

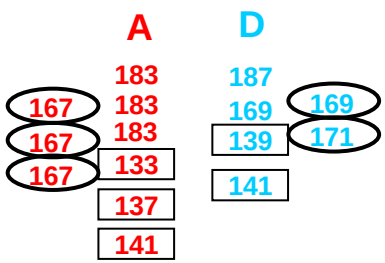


02(Mother)
n=4

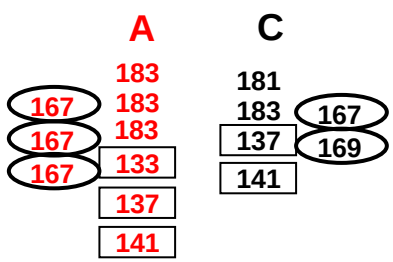


Children

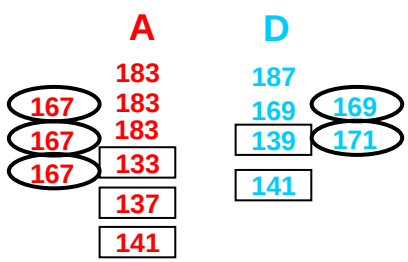
3(n=5)



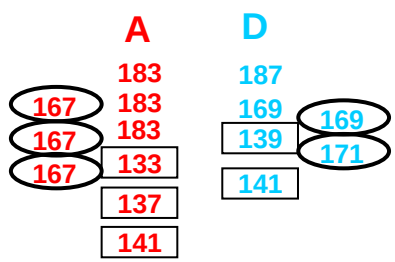
4(n=5)



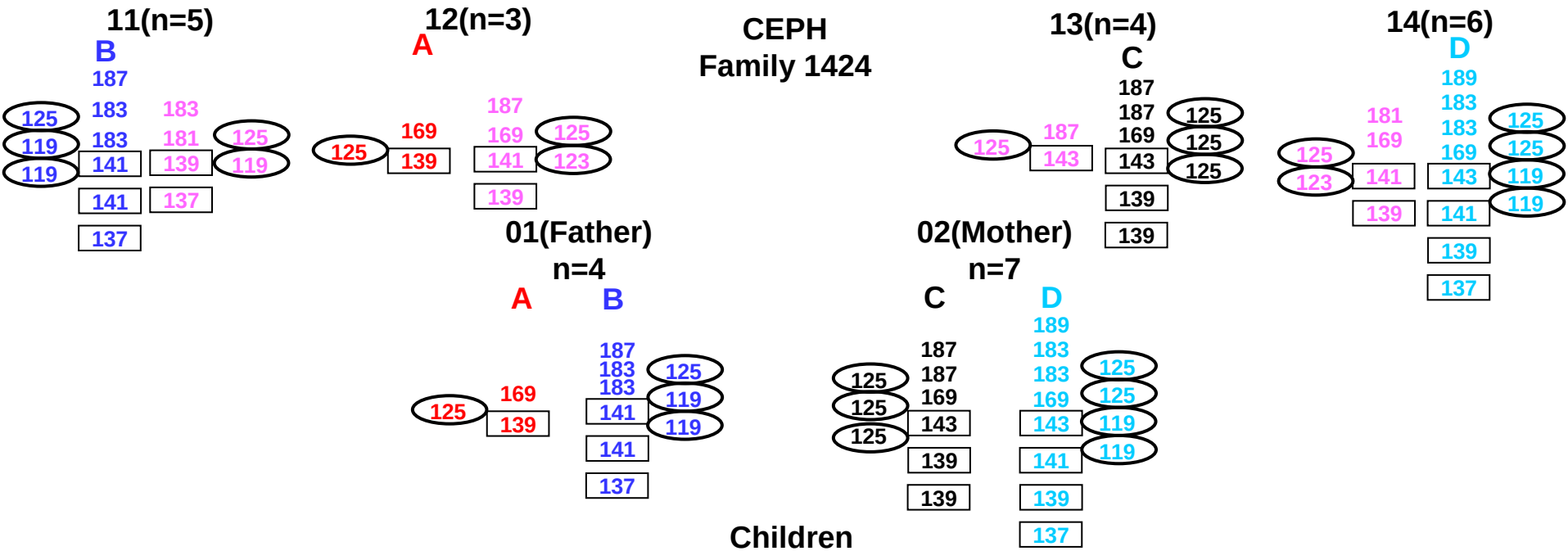
5(n=5)



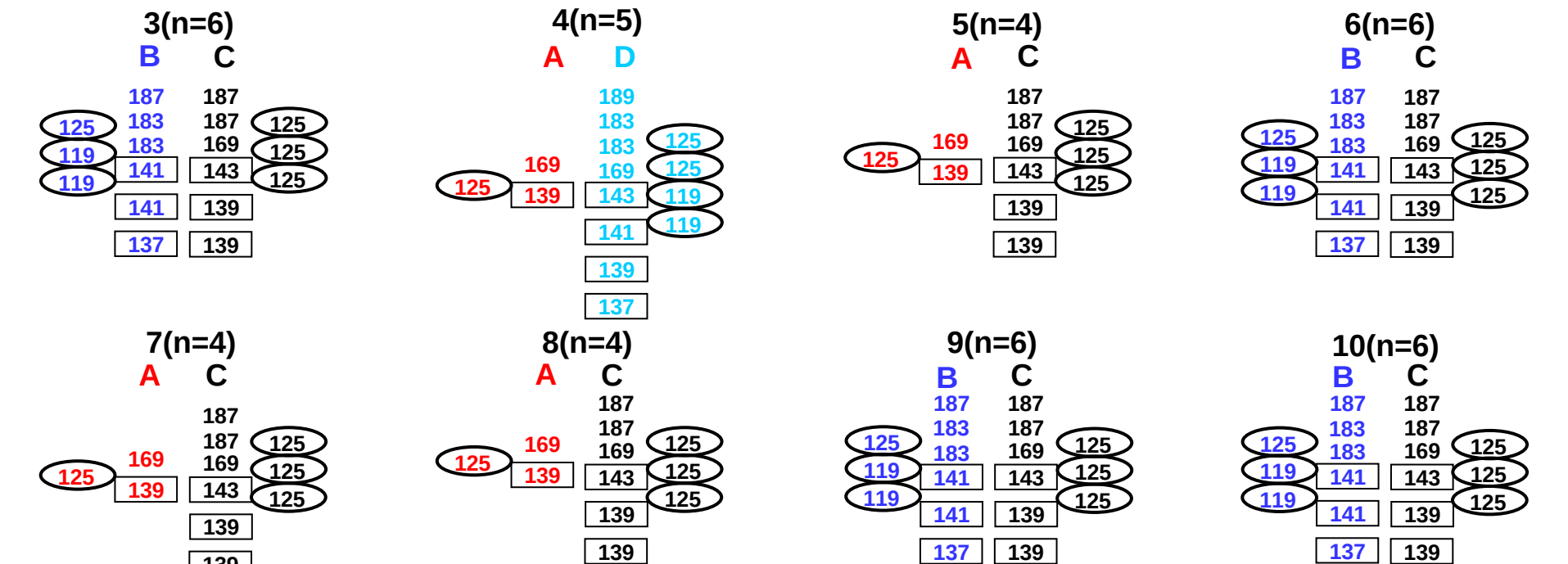
6(n=5)



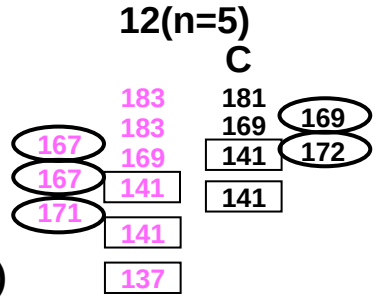
CEPH Family 1424

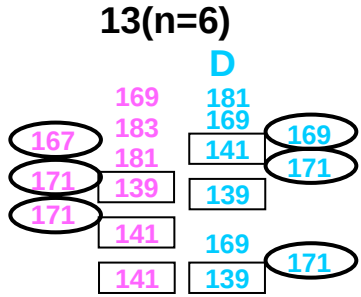


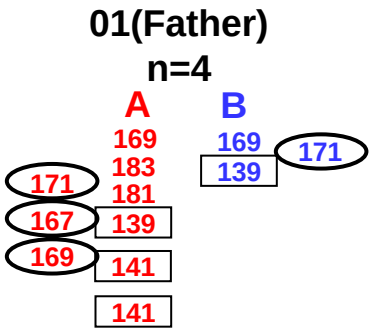
Children

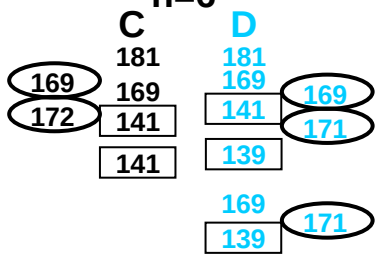


CEPH Family 13292



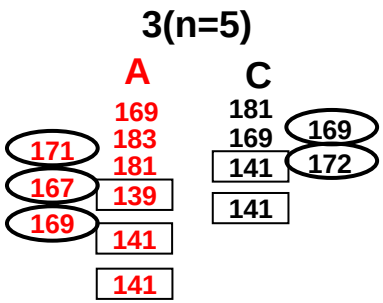


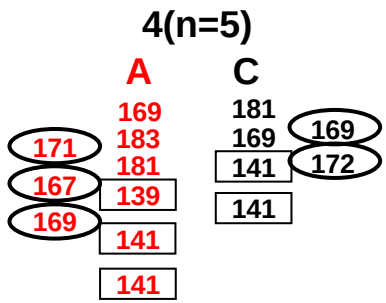


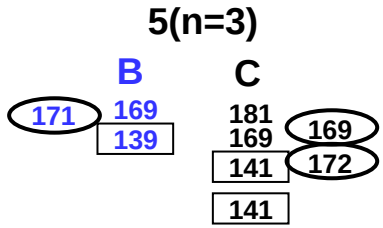


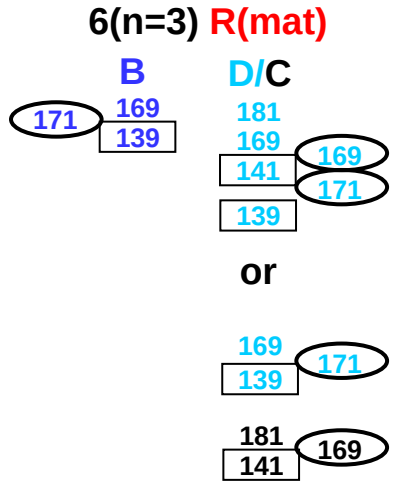
Nature of crossover in child 6, and placement of maternal repeats, ambiguous because of allele sharing between maternal haplotypes

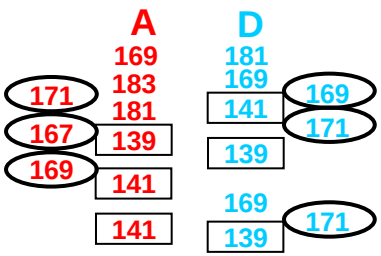
Children

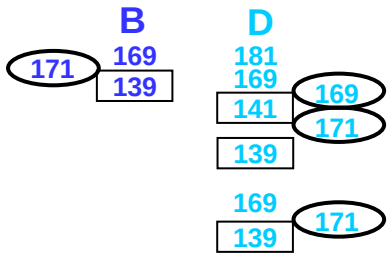


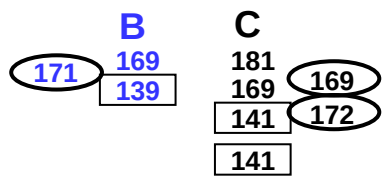




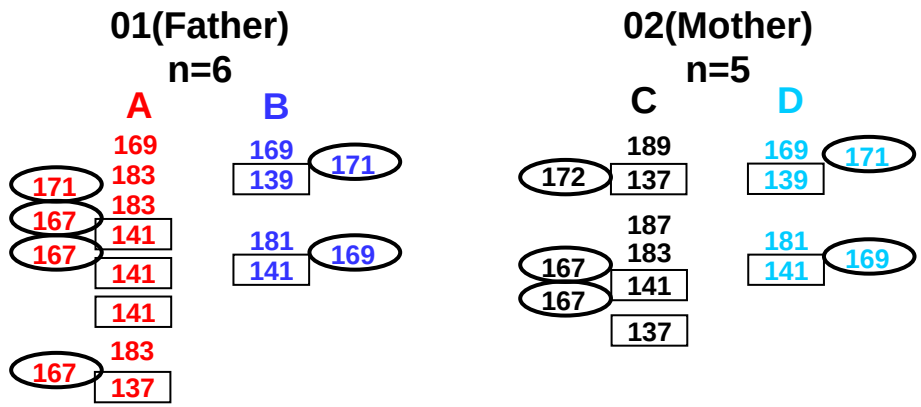








CEPH
Family 13294



Children