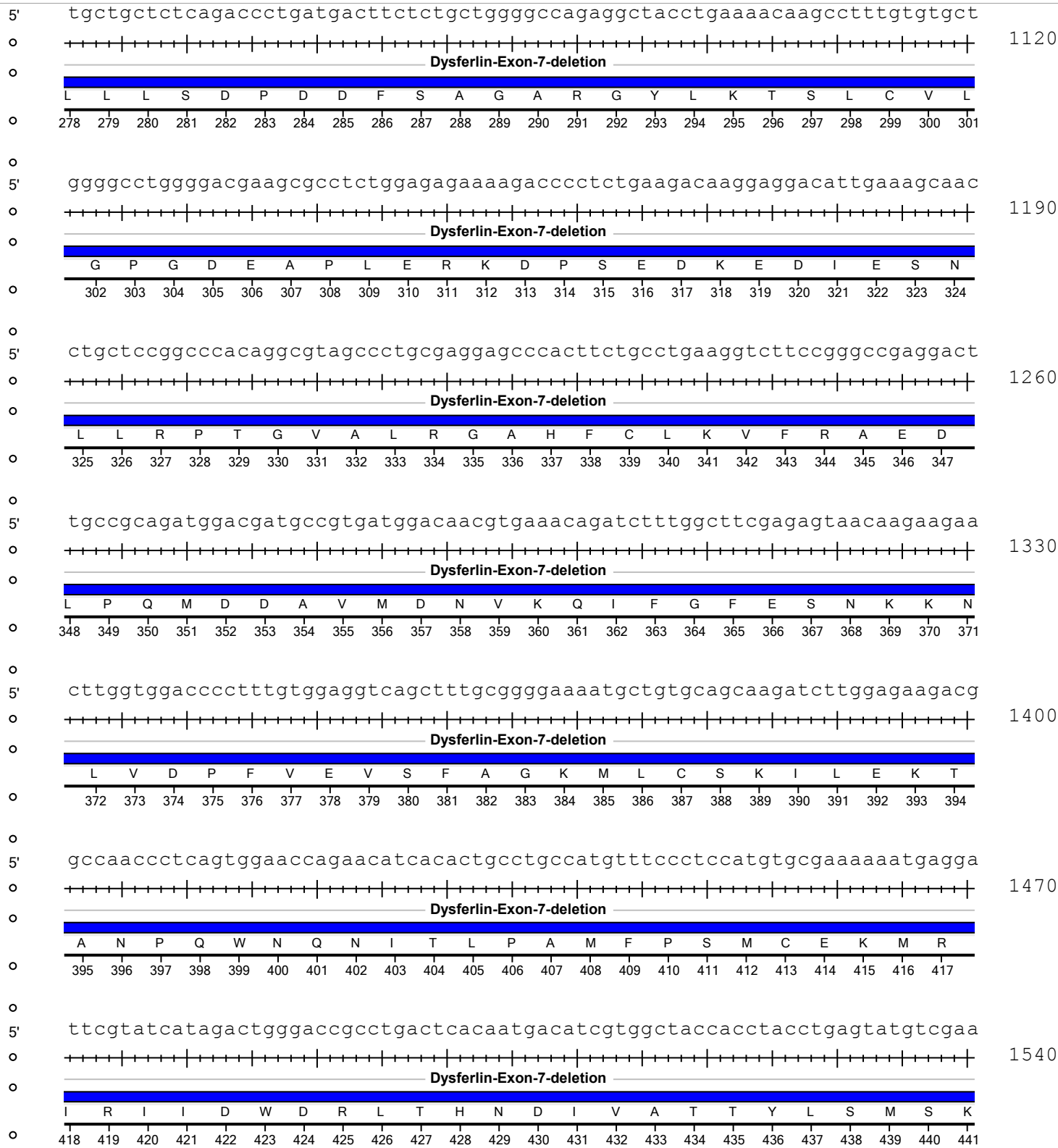


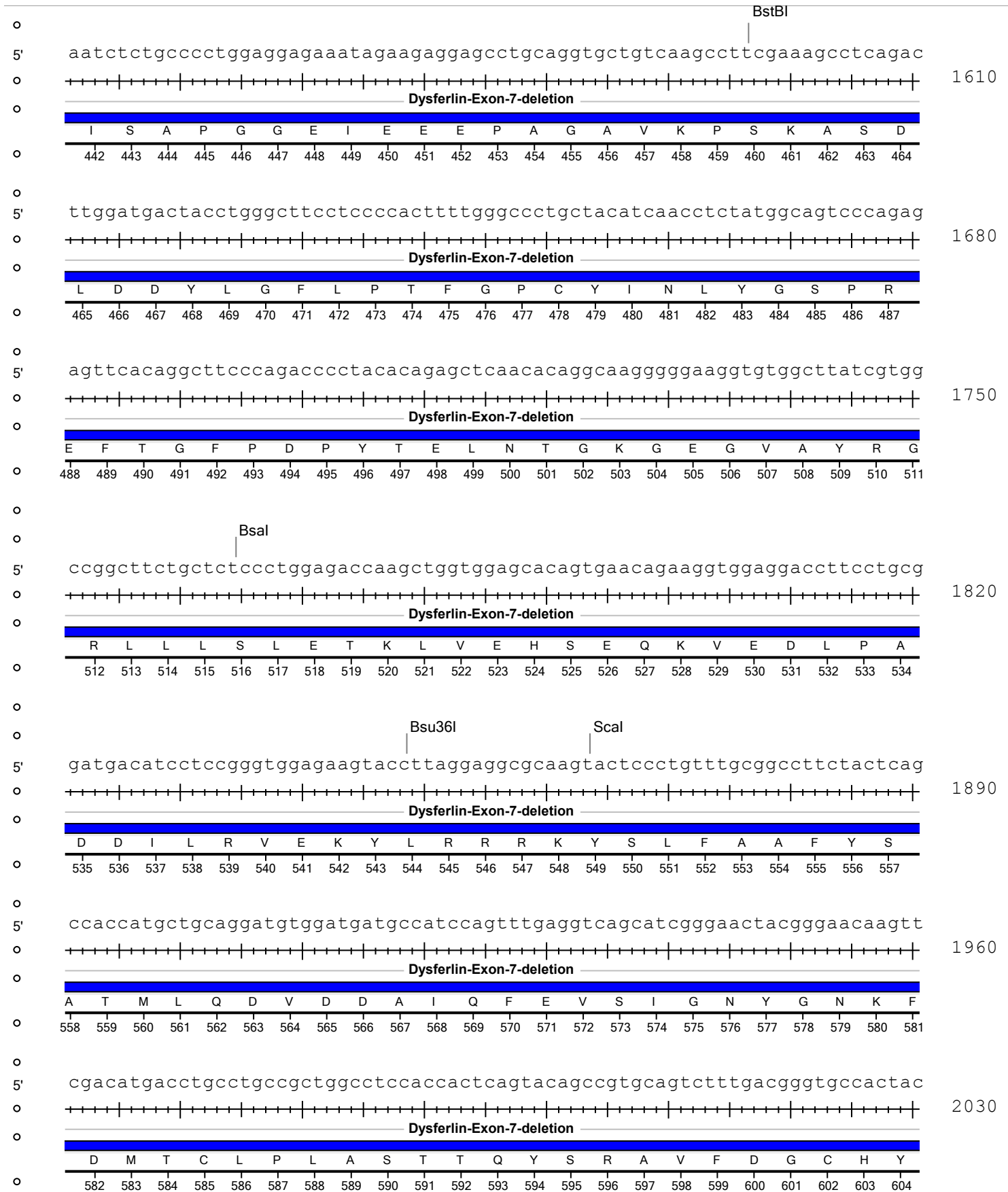
5' tcgcgcggtttcgggtgatgacgggtgaaaacctctgacacatgcagctcccggaagacgggtcacagcttgtct
0 ++++++|+++++|+++++|+++++|+++++|+++++|+++++|+++++|+++++|+++++|+++++|+++++|+++++|+++++|+++++
0
5' gtaagcggatgccgggagcagacaagcccgtcaggggcgcgtcagcgggtgttggcgggtgtcggggctgg
0 ++++++|+++++|+++++|+++++|+++++|+++++|+++++|+++++|+++++|+++++|+++++|+++++|+++++|+++++|+++++
0
0 EcoRI EcoRV HpaI Acc65I KpnI Spel
0 cttaactatgcggcatcagagcagattgtactgagagtgcacgaattcgatatcgtaaaccgtaccacta
0 ++++++|+++++|+++++|+++++|+++++|+++++|+++++|+++++|+++++|+++++|+++++|+++++|+++++|+++++|+++++
0 MluI
0 gtacgcgtatgctgagggctcttcatacctctatgccgagaacgtccacacacccgcacaccgacatcagcga
0 ++++++|+++++|+++++|+++++|+++++|+++++|+++++|+++++|+++++|+++++|+++++|+++++|+++++|+++++|+++++
0 Dysferlin-Exon-7-deletion
0 M L R V F I L Y A E N V H T P D T D I S D
0 1 2 3 4 5 6 7 8 9 10 11 12 13 14 15 16 17 18 19 20 21
0
0 SacII
0 tgctactgctccgcgggtgtttgcaggggtgaagaagagaaccaaagtcatacaagaacagcgtgaacct
0 ++++++|+++++|+++++|+++++|+++++|+++++|+++++|+++++|+++++|+++++|+++++|+++++|+++++|+++++|+++++
0 Dysferlin-Exon-7-deletion
0 A Y C S A V F A G V K K R T K V I K N S V N P
0 22 23 24 25 26 27 28 29 30 31 32 33 34 35 36 37 38 39 40 41 42 43 44
0
5' gtatggaatgagggatttgaatgggacctcaagggcatccccctggaccagggctctgagcttcatgtgg
0 ++++++|+++++|+++++|+++++|+++++|+++++|+++++|+++++|+++++|+++++|+++++|+++++|+++++|+++++|+++++
0 Dysferlin-Exon-7-deletion
0 V W N E G F E W D L K G I P L D Q G S E L H V
0 45 46 47 48 49 50 51 52 53 54 55 56 57 58 59 60 61 62 63 64 65 66 67
0
5' tggtaaagaccatgagacgatggggaggaacaggttcctgggggaagccaagggtcccactccgagaggt
0 ++++++|+++++|+++++|+++++|+++++|+++++|+++++|+++++|+++++|+++++|+++++|+++++|+++++|+++++|+++++
0 Dysferlin-Exon-7-deletion
0 V V K D H E T M G R N R F L G E A K V P L R E V
0 68 69 70 71 72 73 74 75 76 77 78 79 80 81 82 83 84 85 86 87 88 89 90 91
0
5' cctcgccacccttagtgtgtccgccagcttcaatgccccctgctggacaccaagaagcagccacaggg
0 ++++++|+++++|+++++|+++++|+++++|+++++|+++++|+++++|+++++|+++++|+++++|+++++|+++++|+++++|+++++
0 Dysferlin-Exon-7-deletion
0 L A T P S L S A S F N A P L L D T K K Q P T G
0 92 93 94 95 96 97 98 99 100 101 102 103 104 105 106 107 108 109 110 111 112 113 114

5'
 cctcgcgtggtcctcgcaggtgtcctacacacccgctgcctggagctgtgcccctgttcccgccttactc
 0
 0
 Dysferlin-Exon-7-deletion
 A S L V L Q V S Y T P L P G A V P L F P P P T
 115 116 117 118 119 120 121 122 123 124 125 126 127 128 129 130 131 132 133 134 135 136 137
 0
 5'
 ctctggagccctccccgactctgcctgacctggatgtagtggcagacacaggaggagaggaagacacaga
 0
 0
 Dysferlin-Exon-7-deletion
 P L E P S P T L P D L D V V A D T G G E E D T E
 138 139 140 141 142 143 144 145 146 147 148 149 150 151 152 153 154 155 156 157 158 159 160 161
 0
 5'
 ggaccagggactcactggagatgaggcggagccattcctggatcaaagcggaggcccgggggctcccacc
 0
 0
 Dysferlin-Exon-7-deletion
 D Q G L T G D E A E P F L D Q S G G P G A P T
 162 163 164 165 166 167 168 169 170 171 172 173 174 175 176 177 178 179 180 181 182 183 184
 0
 5'
 accccaaggaaactaccttcacgtcctccgccccactacccgggatcaaaagaaagcgaagtgcgccta
 0
 0
 Dysferlin-Exon-7-deletion
 T P R K L P S R P P P H Y P G I K R K R S A P
 185 186 187 188 189 190 191 192 193 194 195 196 197 198 199 200 201 202 203 204 205 206 207
 0
 0
 XbaI
 |
 5'
 catctagaaagctgctgtcagacaaaccgcaggatttccagactcttttcttcaacttgtttgactctcc
 0
 0
 Dysferlin-Exon-7-deletion
 T S R K L L S D K P Q D F Q T L F F N L F D S P
 208 209 210 211 212 213 214 215 216 217 218 219 220 221 222 223 224 225 226 227 228 229 230 231
 0
 0
 Deletion
 tggggagctgtttgatgagcccatctttatcacggtagtagactctcgttctctcaggacagatgctctc
 0
 0
 Dysferlin-Exon-7-deletion
 G E L F D E P I F I T V V D S R S L R T D A L
 232 233 234 235 236 237 238 239 240 241 242 243 244 245 246 247 248 249 250 251 252 253 254
 0
 5'
 ctcggggagttccggatggacgtgggcaccatttacagagagccccggcacgcctatctcaggaagtggc
 0
 0
 Dysferlin-Exon-7-deletion
 L G E F R M D V G T I Y R E P R H A Y L R K W
 255 256 257 258 259 260 261 262 263 264 265 266 267 268 269 270 271 272 273 274 275 276 277

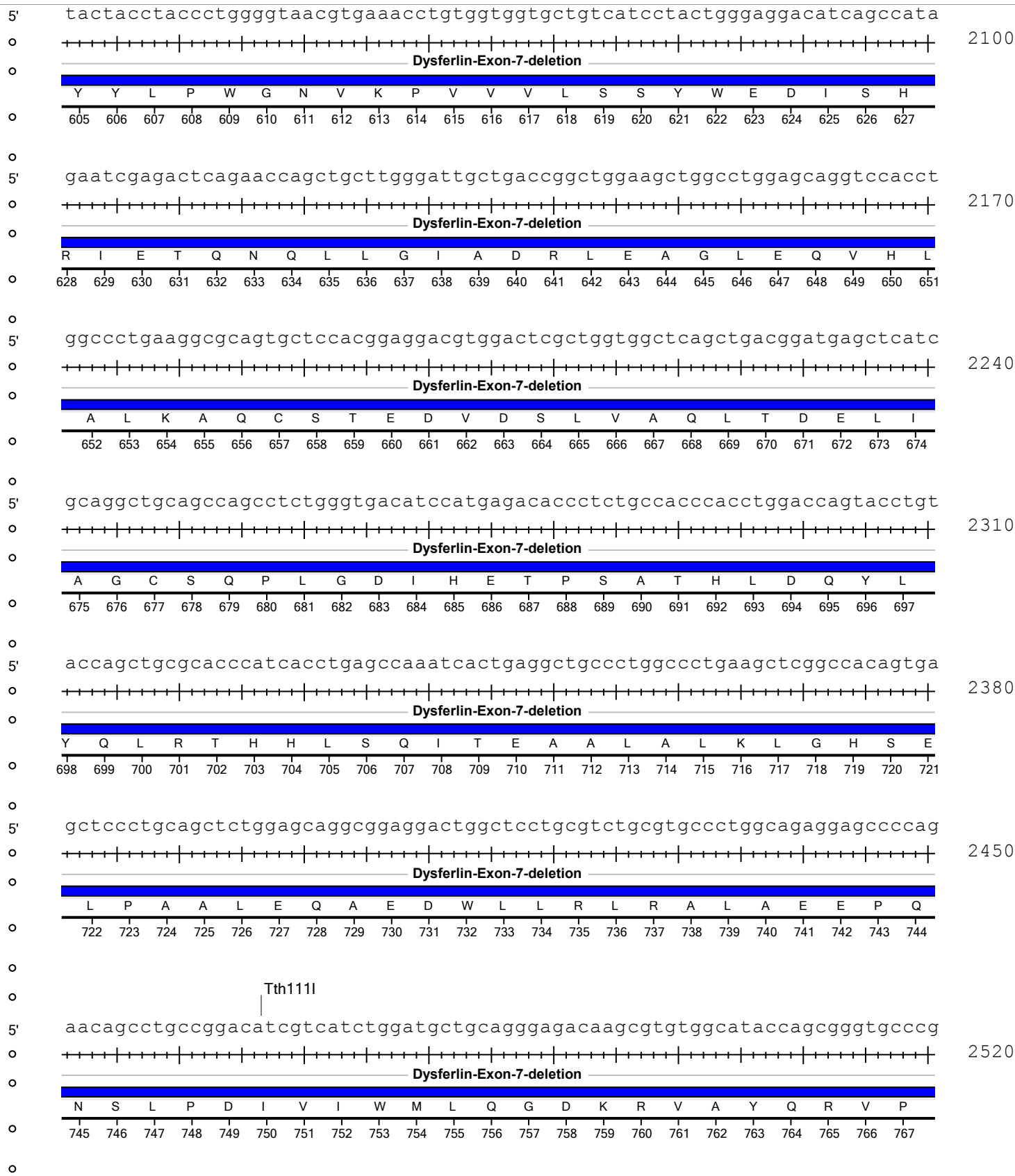
Dysferlin exon 7 deletion in pUC57-Kan



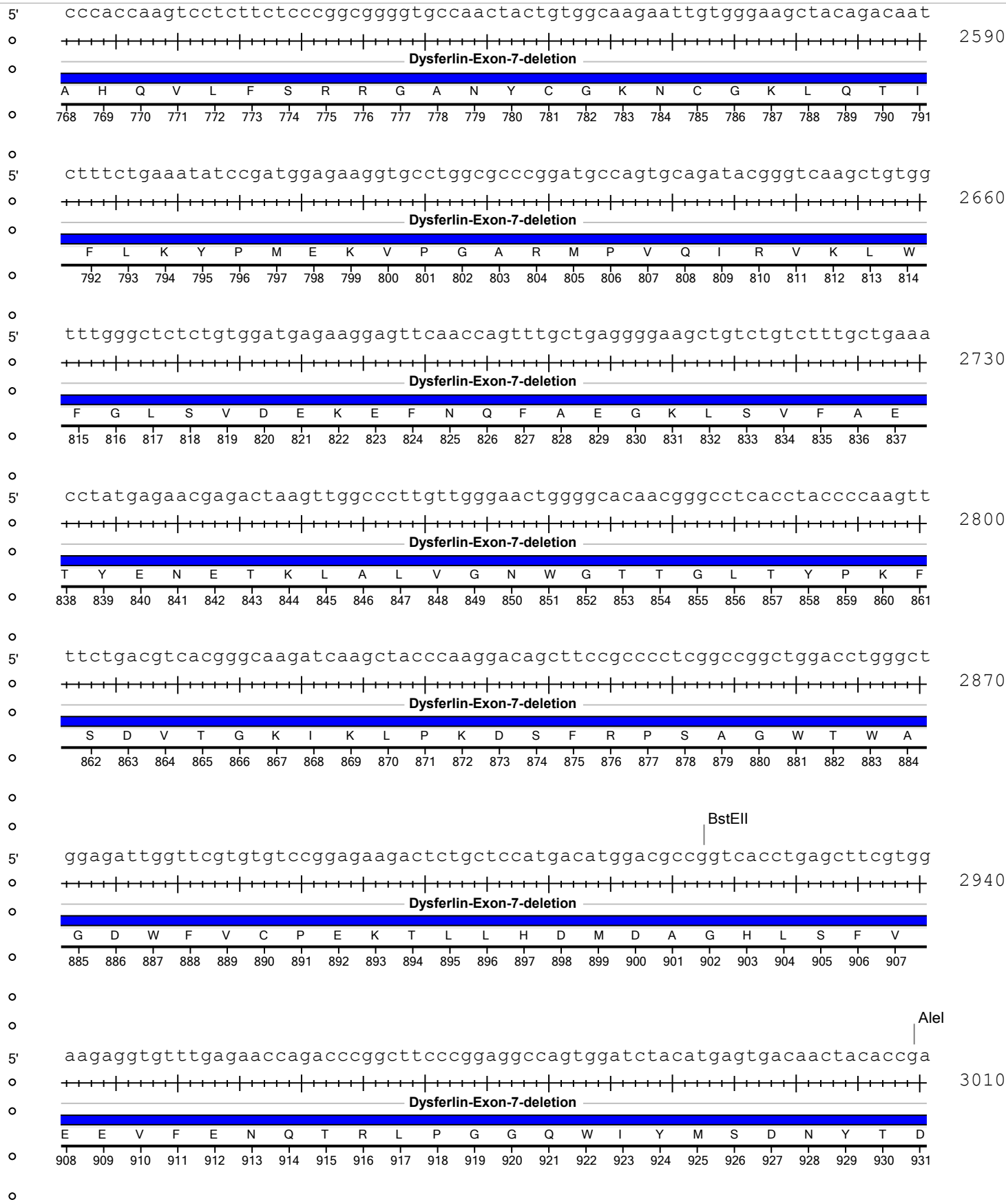
Dysferlin exon 7 deletion in pUC57-Kan



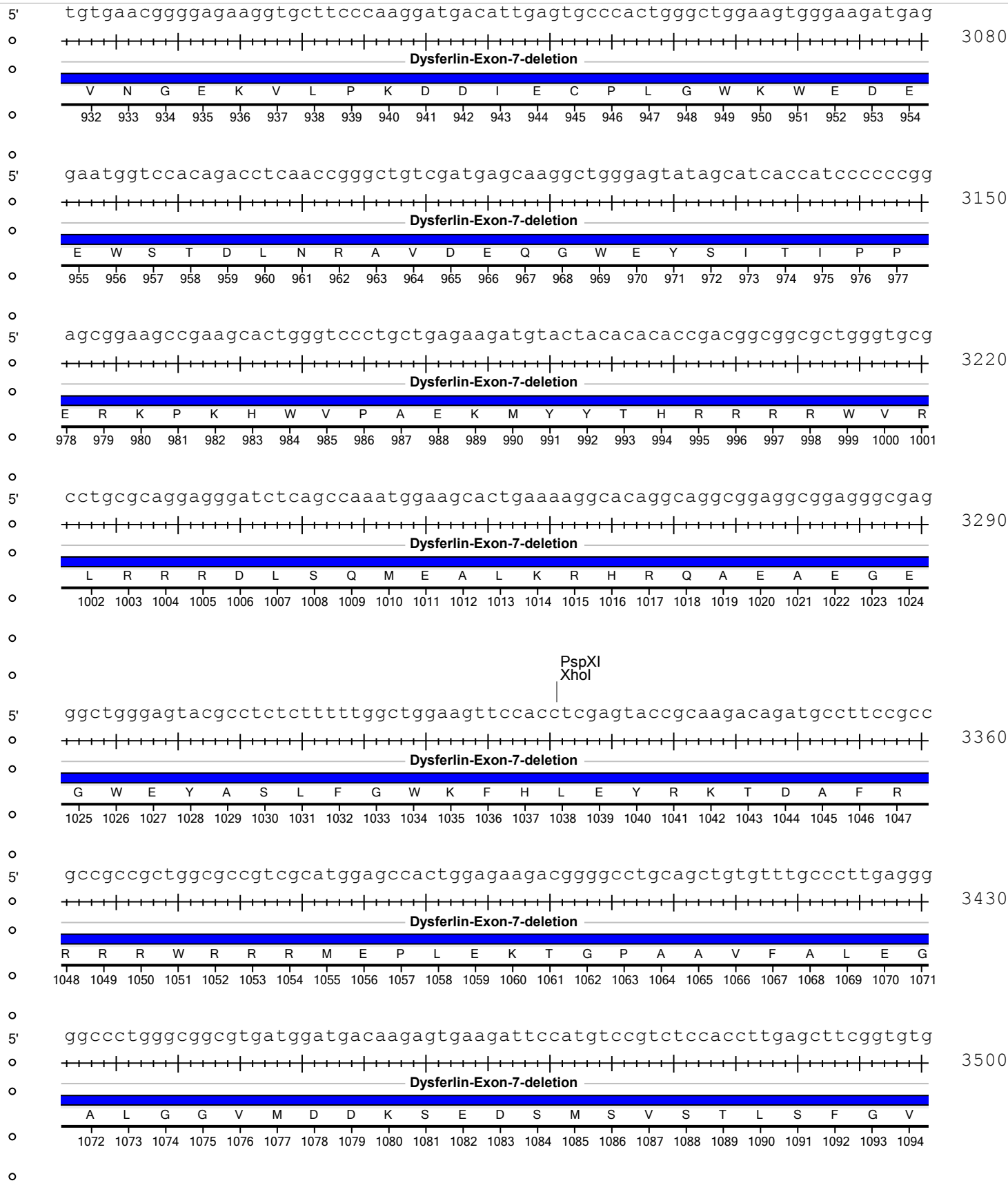
Dysferlin exon 7 deletion in pUC57-Kan



Dysferlin exon 7 deletion in pUC57-Kan



Dysferlin exon 7 deletion in pUC57-Kan



5' aacagaccacgatttcctgcataattcgggaacctaccatctacgctgctacatgtaccag
+
Dysferlin-Exon-7-deletion
N R P T I S C I F D Y G N R Y H L R C Y M Y Q
1095 1096 1097 1098 1099 1100 1101 1102 1103 1104 1105 1106 1107 1108 1109 1110 1111 1112 1113 1114 1115 1116 1117

5' cccgggacctggctgcgatggacaaggactctttttctgatccctatgccatcgctctccttctgcacca
+
Dysferlin-Exon-7-deletion
A R D L A A M D K D S F S D P Y A I V S F L H Q
1118 1119 1120 1121 1122 1123 1124 1125 1126 1127 1128 1129 1130 1131 1132 1133 1134 1135 1136 1137 1138 1139 1140 1141

5' gagccagaagacggtggtggtgaagaacacccttaacccccacctgggaccagacgctcatcttctacgag
+
Dysferlin-Exon-7-deletion
S Q K T V V V K N T L N P T W D Q T L I F Y E
1142 1143 1144 1145 1146 1147 1148 1149 1150 1151 1152 1153 1154 1155 1156 1157 1158 1159 1160 1161 1162 1163 1164

5' atcgagatctttggcgagccggccacagttgctgagcaaccgcccagcattgtggtggagctgtacgacc
+
Dysferlin-Exon-7-deletion
I E I F G E P A T V A E Q P P S I V V E L Y D
1165 1166 1167 1168 1169 1170 1171 1172 1173 1174 1175 1176 1177 1178 1179 1180 1181 1182 1183 1184 1185 1186 1187

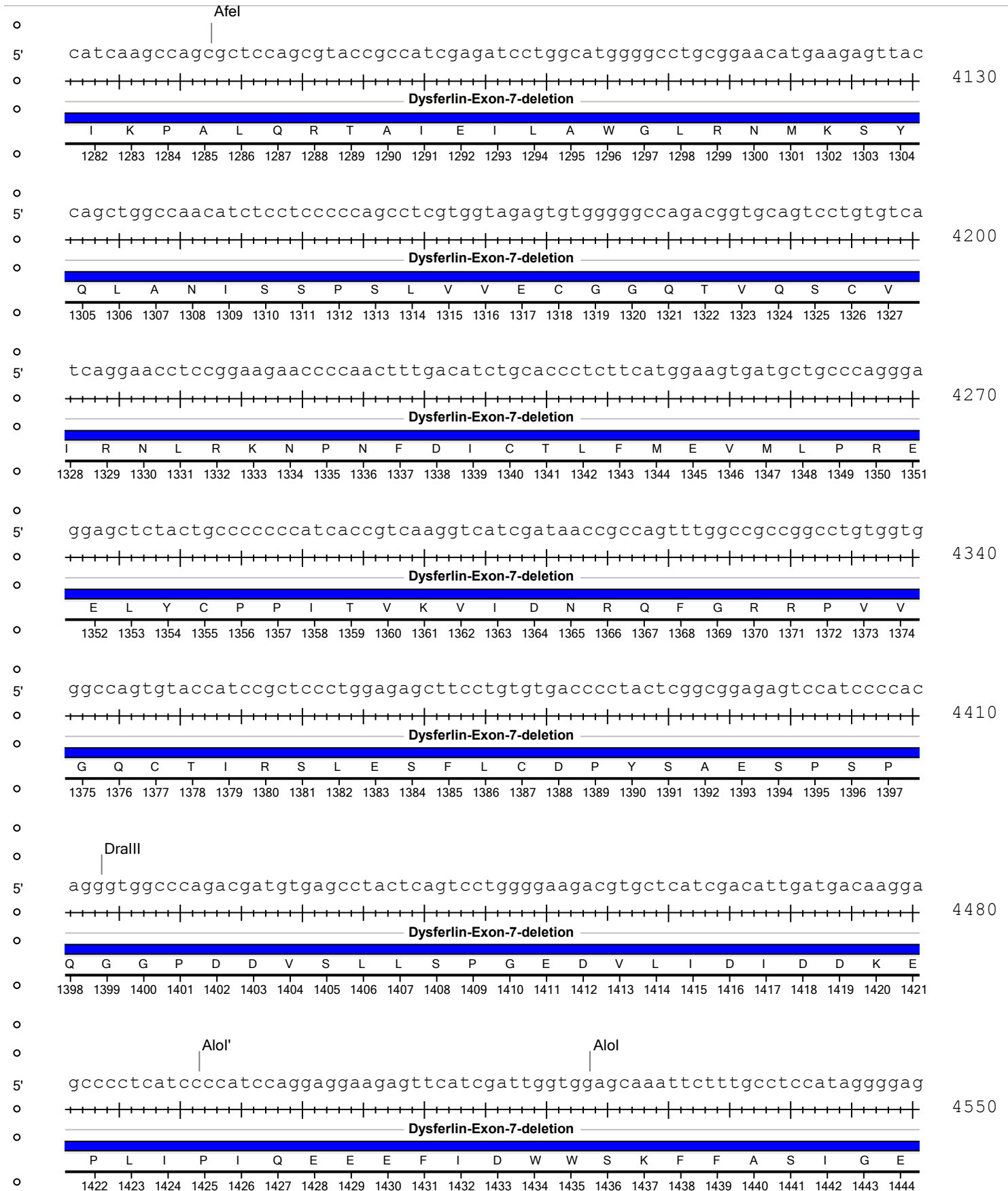
5' atgacacttatggtgcagacgagtttatgggtcgctgcattgtcaaccgagctctggaacggatgccacg
+
Dysferlin-Exon-7-deletion
H D T Y G A D E F M G R C I C Q P S L E R M P R
1188 1189 1190 1191 1192 1193 1194 1195 1196 1197 1198 1199 1200 1201 1202 1203 1204 1205 1206 1207 1208 1209 1210 1211

5' gctggcctggttcccactgacgagggggcagccagccgctcgggggagctgctggcctcttttgagctcatc
+
Dysferlin-Exon-7-deletion
L A W F P L T R G S Q P S G E L L A S F E L I
1212 1213 1214 1215 1216 1217 1218 1219 1220 1221 1222 1223 1224 1225 1226 1227 1228 1229 1230 1231 1232 1233 1234

5' cagagagagaagccggccatccaccatattcctggttttgaggtgcaggagacatcaaggatcctggatg
+
Dysferlin-Exon-7-deletion
Q R E K P A I H H I P G F E V Q E T S R I L D
1235 1236 1237 1238 1239 1240 1241 1242 1243 1244 1245 1246 1247 1248 1249 1250 1251 1252 1253 1254 1255 1256 1257

5' agtctgaggacacagacctgcctacccaccaccccccagaggaggccaacatctacatggttcctcagaa
+
Dysferlin-Exon-7-deletion
E S E D T D L P Y P P P Q R E A N I Y M V P Q N
1258 1259 1260 1261 1262 1263 1264 1265 1266 1267 1268 1269 1270 1271 1272 1273 1274 1275 1276 1277 1278 1279 1280 1281

Dysferlin exon 7 deletion in pUC57-Kan



5' agggaaaagtgcggtccttaccttgagaaaggattttgacaccctgaaggtcttatgacacacagctggaga
+
Dysferlin-Exon-7-deletion
R E K C G S Y L E K D F D T L K V Y D T Q L E
1445 1446 1447 1448 1449 1450 1451 1452 1453 1454 1455 1456 1457 1458 1459 1460 1461 1462 1463 1464 1465 1466 1467

Stul
atgtggaggccttttgaggcctgtctgacttttctaacaaccttcaggctgtaccggggcaagacgcagga
+
Dysferlin-Exon-7-deletion
N V E A F E G L S D F C N T F K L Y R G K T Q E
1468 1469 1470 1471 1472 1473 1474 1475 1476 1477 1478 1479 1480 1481 1482 1483 1484 1485 1486 1487 1488 1489 1490 1491

ggagacagaagatccatctgtgattggtgaatttaaggcgctcttcaaaaatttatcccctcccagaagac
+
Dysferlin-Exon-7-deletion
E T E D P S V I G E F K G L F K I Y P L P E D
1492 1493 1494 1495 1496 1497 1498 1499 1500 1501 1502 1503 1504 1505 1506 1507 1508 1509 1510 1511 1512 1513 1514

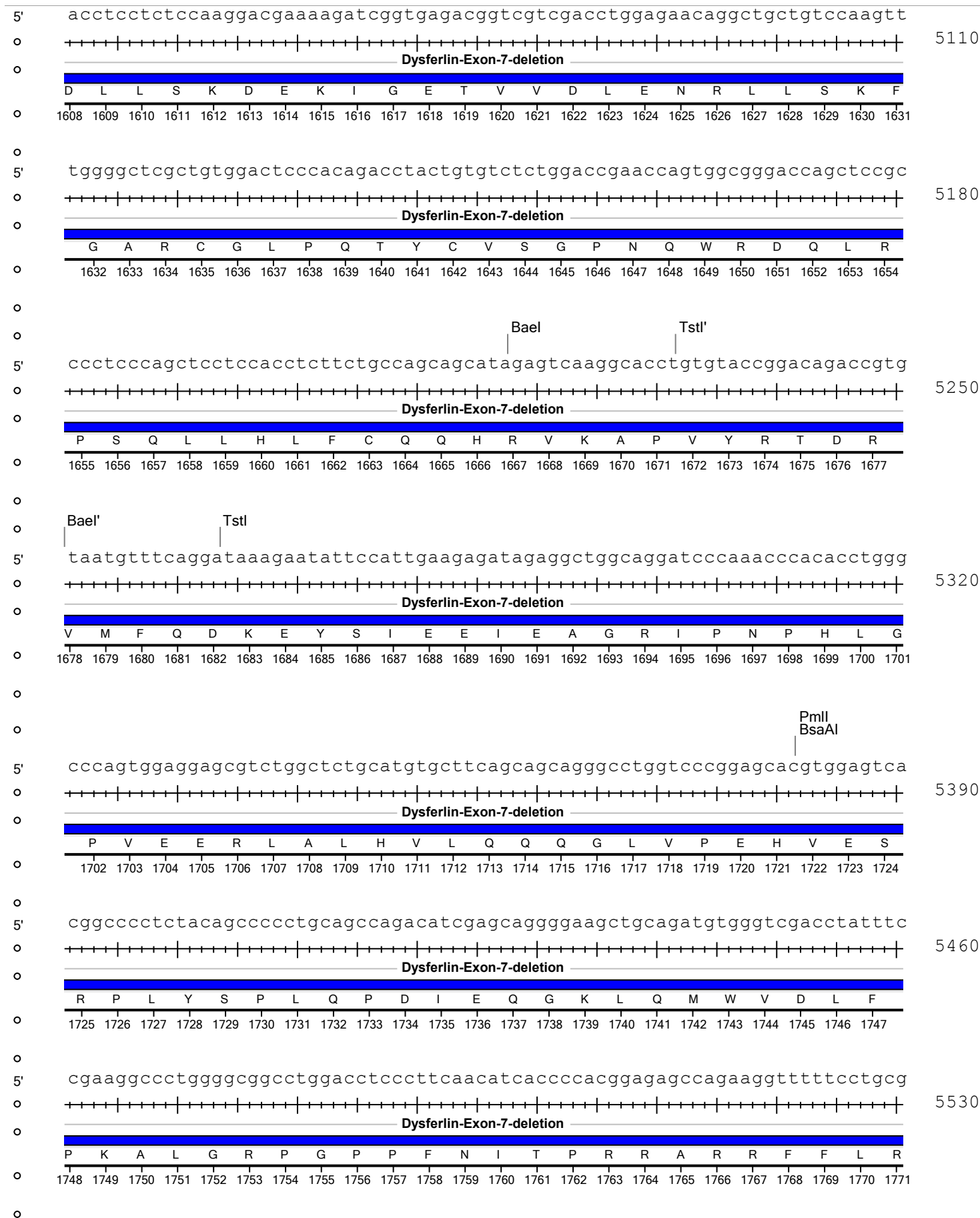
ccagccatccccatgcccccaagacagttccaccagctggccgccccaggagaccccaggagtgcttggtcc
+
Dysferlin-Exon-7-deletion
P A I P M P P R Q F H Q L A A Q G P Q E C L V
1515 1516 1517 1518 1519 1520 1521 1522 1523 1524 1525 1526 1527 1528 1529 1530 1531 1532 1533 1534 1535 1536 1537

gtatctacattgtccgagcatttggcctgcagcccaaggaccccaatggaaagtgtgatccttacatcaa
+
Dysferlin-Exon-7-deletion
R I Y I V R A F G L Q P K D P N G K C D P Y I K
1538 1539 1540 1541 1542 1543 1544 1545 1546 1547 1548 1549 1550 1551 1552 1553 1554 1555 1556 1557 1558 1559 1560 1561

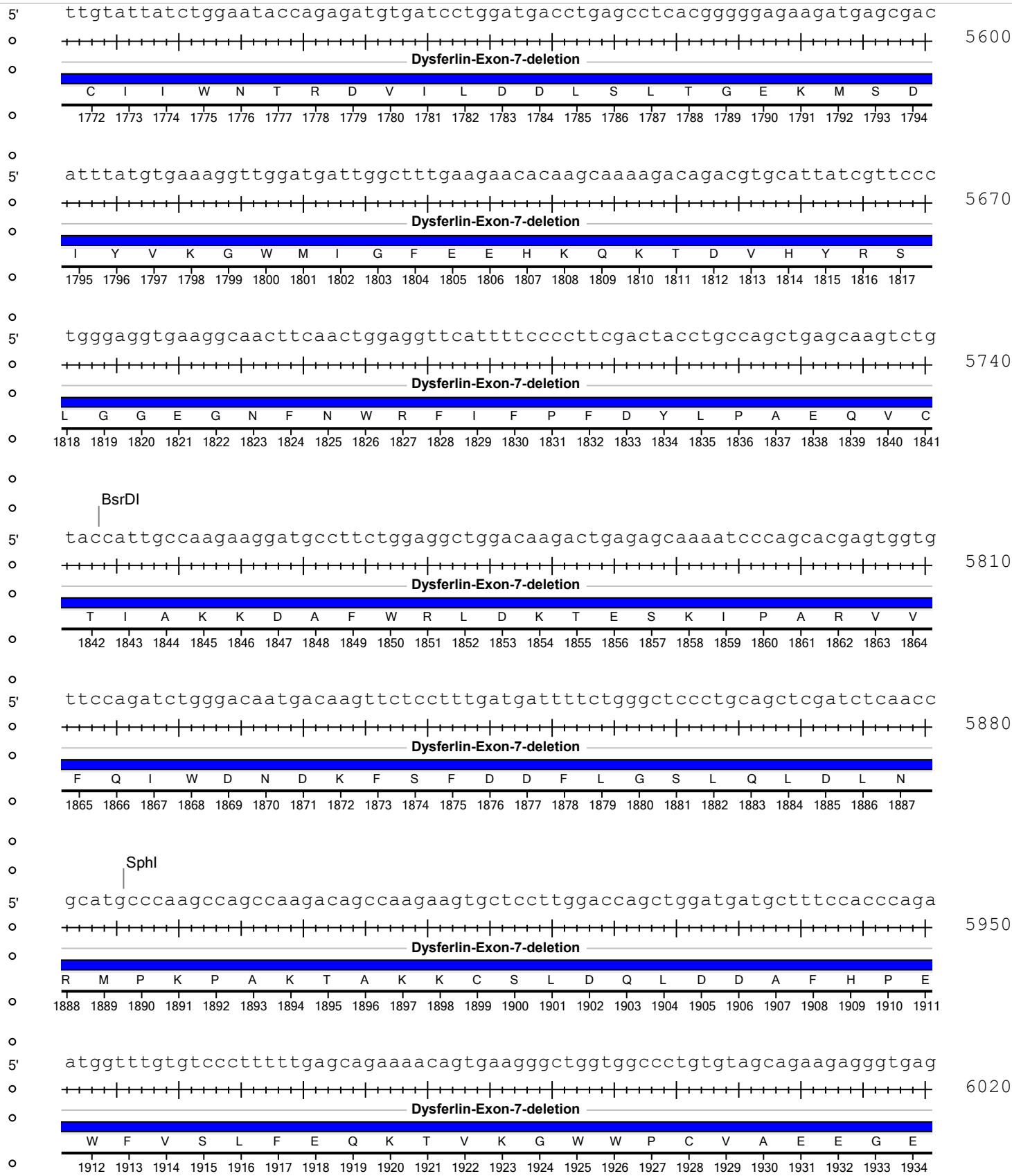
gatctccataggggaagaaatcagtgagtgaccaggataactacatcccctgcacgctggagcccgtattt
+
Dysferlin-Exon-7-deletion
I S I G K K S V S D Q D N Y I P C T L E P V F
1562 1563 1564 1565 1566 1567 1568 1569 1570 1571 1572 1573 1574 1575 1576 1577 1578 1579 1580 1581 1582 1583 1584

ggaaagatgttcgagctgacctgcactctgcctctggagaaggacctaagatcactctctatgactatg
+
Dysferlin-Exon-7-deletion
G K M F E L T C T L P L E K D L K I T L Y D Y
1585 1586 1587 1588 1589 1590 1591 1592 1593 1594 1595 1596 1597 1598 1599 1600 1601 1602 1603 1604 1605 1606 1607

Dysferlin exon 7 deletion in pUC57-Kan



Dysferlin exon 7 deletion in pUC57-Kan



5' aagaaaaatactggcgggcaagctggaaatgaccttggagattgtagcagagagtgagcatgaggagcggc

o

o

Dysferlin-Exon-7-deletion

K K I L A G K L E M T L E I V A E S E H E E R

1935 1936 1937 1938 1939 1940 1941 1942 1943 1944 1945 1946 1947 1948 1949 1950 1951 1952 1953 1954 1955 1956 1957

Genomic map of the Dysferlin gene region. The map shows the DNA sequence: 5' ctgctggccagggccgggatgagcccaacatgaaccctaagcttgaggacccaaggcgccccgacacctc. A HindIII restriction site is indicated by a vertical line above the sequence. A blue bar below the sequence represents the Dysferlin-Exon-7-deletion. The timeline at the bottom shows years from 1958 to 1981, with corresponding amino acid positions: P (1958), A (1959), G (1960), Q (1961), G (1962), R (1963), D (1964), E (1965), P (1966), N (1967), M (1968), N (1969), P (1970), K (1971), L (1972), E (1973), D (1974), P (1975), R (1976), R (1977), P (1978), D (1979), T (1980), S (1981).

Genomic map of the Dysferlin gene on chromosome 2p11.2. The map shows the gene structure with exons represented by blue boxes and introns by lines with arrows. A red box highlights the 'Dysferlin-Exon-7-deletion' region. The map includes a scale bar from 1982 to 2004, with markers for F, L, W, F, T, S, P, Y, K, T, M, K, F, I, L, W, R, R, F, R, W, A, I.

5' atcctcttcatcctcttcatcctgctgctgttccctggccatcttcatctacgccttcccgaactatg

Dysferlin-Exon-7-deletion

2005 2006 2007 2008 2009 2010 2011 2012 2013 2014 2015 2016 2017 2018 2019 2020 2021 2022 2023 2024 2025 2026 2027

Genomic map of the Dysferlin gene (DYSF) on chromosome 2p13. The map shows the gene structure with 11 exons (numbered 1-11) and 10 introns. A blue arrow indicates the location of the Dysferlin-Exon-7-deletion, spanning from the start of Exon 7 to the end of Exon 8. Restriction enzyme sites are marked above the gene: BclI, NdeI, NheI, BmtI, and NotI. The DNA sequence for Exon 7 is shown: ctgccatgaagctggtgaagcccttcagctgatgatcaccatggcatatgggctagcgcggccgcatggt. The map is aligned with the human genome coordinates from 2028 to 2039.

5' catagctgtttcctgtgtgaaattgttatccgctcacaaattccacacaacatacgagccggaagcataaa
0

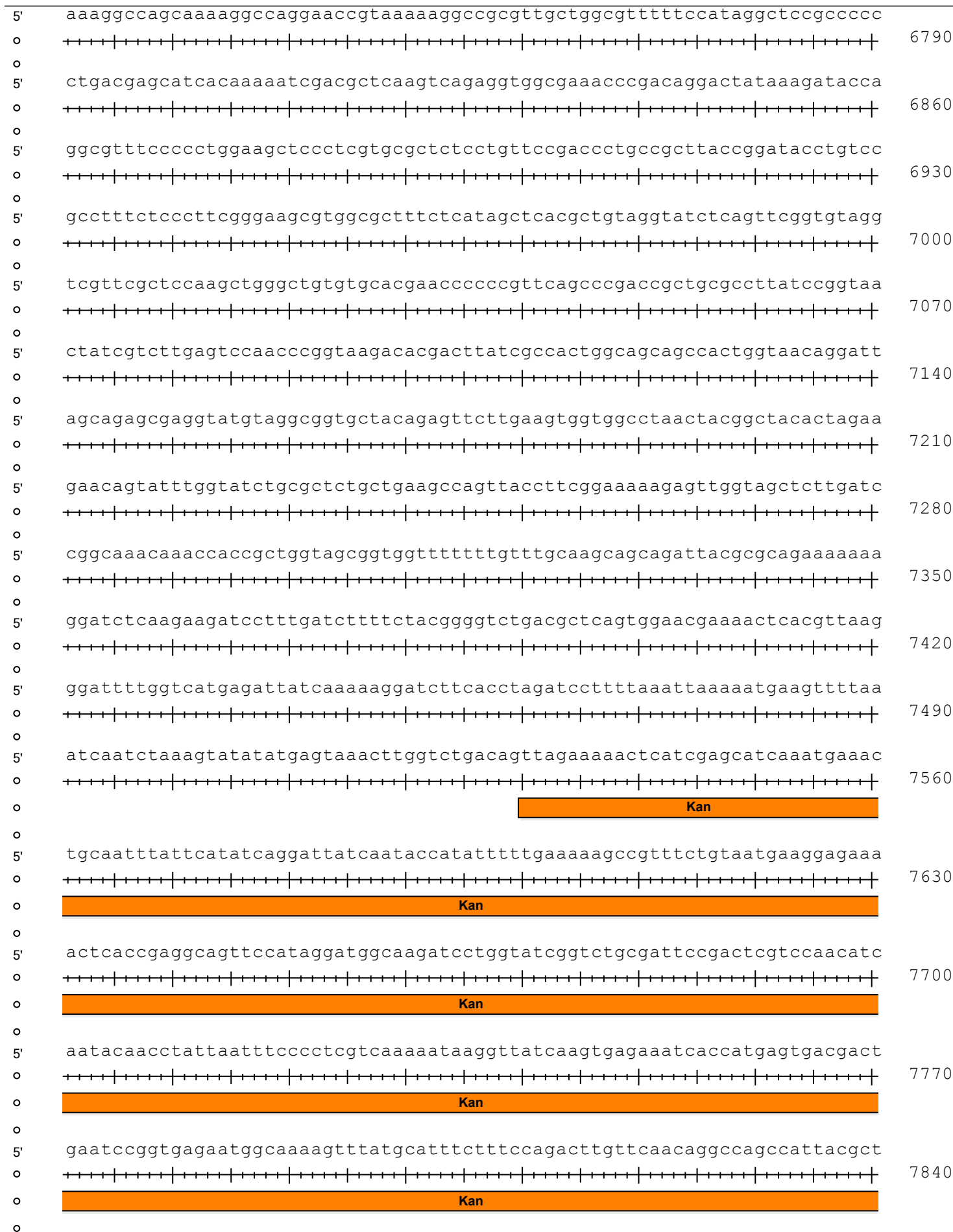
5' gtgtaaagcctggggtgcctaagtgagtgagctaactcacattaattgcgttgcgctcactgcccgctttc
0

5' cagtcgggaaacctgtcgtgccagctgcattaatgaatcggccaacgcgcggggagaggcggtttgcgta
0

3' SapI
 5' ttgggcgctcttcgcgttcctcgctcactgactcgctgcgctcggtcggttcggctgcggcgagcggtatc
 3'

5' agctcactcaaaggcggtacggttatccacagaatcaggggataacgcaggaaagaacatgtgagca
0

Dysferlin exon 7 deletion in pUC57-Kan



Dysferlin exon 7 deletion in pUC57-Kan

