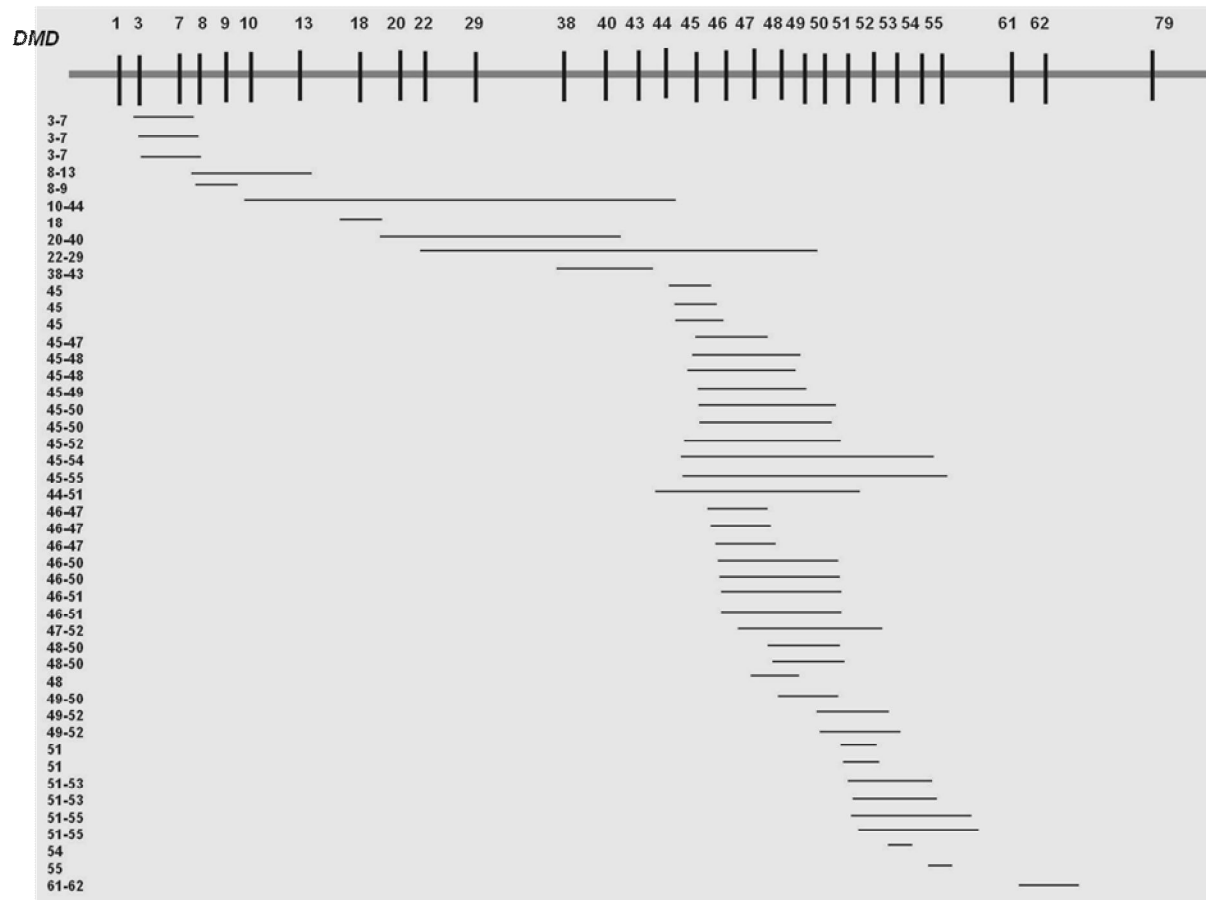


Supplementary Data S1. *Distribution of intragenic deletions across DMD gene*



Supplementary Data S1. Distribution of intragenic deletions across the *DMD* gene as observed in the patient samples analyzed for breakpoint junction sequences (figure not to scale). The top horizontal thick bar represents the *DMD* gene with exons represented as short vertical bars and numbered. The thin horizontal bars represent the region of the gene deleted in each sample.

Supplementary Data S2. Comparison of the deletion breakpoint coordinates

Patient case	Deletion size	5' Breakpoint coordinates			3' Breakpoint coordinates		
		By arrayCGH	By PCR Mapping	Difference (bps)	By arrayCGH	By PCR Mapping	Difference (bps)
4215	114309	32521250	32520801	449	32633250	32635110	1860
1148	518611	32,090,750	32090738	12	32,607,250	32609349	2099
5078	15565	32444250	32443834	416	32455250	32459399	4149
5904	329912	32089250	32090096	846	32419250	32420008	758
2707	118024	32165250	32161987	3263	32279750	32280011	261
7840	176373	31838750	31838786	36	32014750	32015159	409
8094	142792	31797750	31797197	553	31939750	31939989	239
3973	247313	31783250	31782892	358	32029250	32030205	955
3308	66399	31860750	31859830	920	31925750	31926229	479
8120	126575	31874250	31874231	19	32000750	32000806	56
4843	266086	31703750	31703639	111	31969750	31969725	25
6569	411380	31578750	31578698	52	31989750	31990078	328
4325	149308	31734750	31734303	447	31883250	31883611	361
5062	57145	31804300	31804249	51	31860300	31861394	1094
5225	33085	31838750	31838433	317	31871250	31871518	268
4505	33077	31838750	31838441	309	31871250	31871518	268
0454	203169	31667750	31667790	40	31870750	31870959	209
4468	203776	31657250	31657141	109	31860850	31860917	67
6194	149227	31703750	31703527	223	31852250	31852754	504
6730	85336	31741750	31741254	496	31824250	31826590	2340
6224	163908	31628250	31627550	700	31789750	31791458	1708

6215	129587	31573250	31573057	193	31702250	31702644	394
8113	129423	31607250	31607129	121	31736250	31736552	302
0845	30834	31,681,250	31680768	482	31,711,250	31711602	352
4488	113150	31533250	31532833	417	31645750	31645983	233
5024	115639	32674500	32674634	134	32791700	32790273	1427
5510	215285	32630250	32629863	387	32845250	32845148	102
7952	176682	32648250	32648069	181	32824750	32824751	1
9250	71246	32619749	32619772	23	32690749	32691018	269
5366	24400	32379750	32379139	611	32403250	32403539	289
0020	205629	31680249	31679883	366	31885249	31885512	263
2500	252264	31479749	31479771	22	31731249	31732035	786
0837	58366	31709749	31709466	283	31767249	31767832	583
9061	54495	31883749	31884173	424	31938749	31938668	81
9723	23017	31685750	31685409	341	31707750	31708426	676
1094	158381	31588099	31588026	73	31746099	31746407	308
2683	200312	31548750	31546505	2245	31744750	31746817	2067
1154	138602	31210749	31210661	88	31349249	31349263	14
0086	147972	31622750	31617538	5212	31764750	31765510	760
4528	355139	31612749	31612334	415	31967249	31967473	224
4210	456494	31540749	31540317	432	31996749	31996811	62
4297	33200	31743749	31743166	583	31776249	31776366	117
6409	81684	31776249	31775810	439	31857249	31857494	245
0340	59064	32352750	32352586	164	32410750	32411650	900
7573	496490	31680250	31680021	229	32175750	32176511	761
1314	206892	31675249	31674737	512	31881249	31881629	380

2326	212821	31760249	31759828	421	31972249	31972649	400
Average				507			655

Supplementary Data S2. Comparison of the deletion breakpoint coordinates determined individually by array CGH and PCR mapping in each patient case. PCR primer pairs were designed based on the approximate breakpoint coordinates determined by array CGH and exact breakpoints were mapped by sequencing of PCR products. The difference in the breakpoint coordinates determined by each method are shown for both 5' and 3' ends of the deletion and the average of the differences in the coordinates are calculated at the end of the table.

Supplementary Data S3. Breakpoint junction sequence alignment and microhomology determination.

Sample: 0454 Deletion: 46-50

```
5' seq: TAGGTAACAAACCTGCACGTTCTGCACATgtatcccagaacttaaagtaa
      |||
      TAGGTAACAAACCTGCACGTTCTGCACATGAAGAACAAAGTGTGTGTGTA
      |||
3' seq: ttcatagtagtctgaggagattttccatGAAGAACAAAGTGTGTGTGTA
```

Sample: 8113 Deletion: 51-53

```
5' seq: AAATTAGCCAGGCGTGGTGGTACACGctggctagtagtcccagctacta
      |||
      AAATTAGCCAGGCGTGGTGGTACACGCCTGTTGGGGTTGCAACACATGCT
      |||
3' seq: taactagaatcttttcattacttctccCTGTTGGGGTTGCAACACATGCT
```

Sample: 5062 Deletion: 46-47

```
5' seq: GCTACGACATAGATATTATatgatcccactatcacacatgggtgatgac
      |||
      GCTACGACATAGATATTATTAAGTATTTAAATACTTAACATATGTTGT
      |||
3' seq: cttatttgaggaaagagaaaAACTGATTTAAATACTTAACATATGTTGT
```

Sample: 6215 Deletion: 54

```
5' seq: TTTATAGGTTTTGTGTGGGAAAaaatcctcaaaattatgagttacttcc
      |||
      TTTATAGGTTTTGTGTGGGAAAaccAAACAAGATGGGATAAGATCTTCTT
      |||
3' seq: ttttattcaagaaaaaacaaggcaAAACAAGATGGGATAAGATCTTCTT
```

Sample: 4215 Deletion: 8-13

```
5' seq: CGGAAAACGTCTCGGGTCTGGAACCCgaggaggttcggtcccaacaccgt
      |||
      CGGAAAACGTCTCGGGTCTGGAACCCTGTCTCGTTCTGAGGCAGAGCCTC
      |||
```

3' seq: tgcggtgacatgaggtcggacccttTGTCTCGTTCTGAGGCAGAGCCTCC

Sample: 4488 Deletion: 55

5' seq: GTTATCATCAAGTTTGGTTCCTGTCAgcctccagcgactgtaggtggtgc
|||||
GTTATCATCAAGTTTGGTTCCTGTCAAGAAAGAGAAAAATCTGACTCCAAT
|||||
3' seq: gggcaacagttccatggttcctgtatGAAAGAGAAAAATCTGACTCCAAT

Sample: 5510 Deletion: 3-7

5' seq: CATTATTTTTATTGCCCCCAaaaatttcaaacattgtaataaatttca
|||||
CATTATTTTTATTGCCCCCAAGATAGTGAGTGAGTTcCACGAGATCTG
|||||
3' seq: atttcctcagacttttcttgtGATAGTGAGTGAGTTcCACGAGATCTG

Sample: 7952 Deletion: 3-7

5' seq: TattAAAgTAAATATATTAagttaaataaaatacacagaacttta
|| ||| || |||
TAnnAAAnnnAnAnTAnATATATTAACCTAAACnTGTCTcTTTTACAnTGA
|||||
3' seq: ttaccaagttaaaatttgagcaaaCTAAACaTGTCTCaTTTTACAaTGA

Sample: 6569 Deletion: 45-54

5' seq: AGAATACTTCAAGATTACGAACAGGGAGAGGAttatttggttggttacttggtcgttt
|||||
AGAATACTTCAAGATTACGAACAGGGAGAGGATTCTGAATCTTTAGACTCCACAGTCAA
|||||
3' seq: tataatctagttacaaagagtacgactaaagaggtccaGAATCTTTAGACTCCACAGTCAA

Sample: 3688 Deletion: 45-50

5' seq: TTTAGTTTCTATTTTGGACCTtaatatatgtaaactgtataacttaga
|||||
TTTAGTTTCTATTTTGGACCTTAAAGTTCATAGGATTGTCAATTTTGT
|||||
3' seq: cgactcaaagaaatgggaaaccTAAAGTTCATAGGATTGTCAATTTTGT

Sample: 8120 Deletion: 45

5' seq:GGTGGTTGTAACAGTGAGTCCCTCTgagtaagtaacattgaaattgaaat
|||||
GGTGGTTGTAACAGTGAGTCCCTCTGAAAAAAAAATCACTCCAGGTACAGC
|||||
3' seq:ttttgatcatcttaggtagcctgggGAAAAAAAAATCACTCCAGGTACAGC

Sample: 4468

5' seq:TACGTGTCGCAGATCAGTCTACccagaatggtttcaaggacgggtgggat
|||||
TACGTGTCGCAGATCAGTCTACCCAAACCTTTTAAGAAACGACACTCGTC
|||||
3' seq:tgaaatcgtaatgattgtaaatcCAAACCTTTTAAGAAACGACACTCGTC

Sample: 4843

5' seq:TTTTAATAACCTGATACATCAAACcttactttccaaagttcctttcgacg
|||||
TTTTAATAACCTGATACATCAAACACTATCGGGAGCATGTACGTAGGTG
|||||
3' seq:gaactcgtagtagagatgacagttTACTATCGGGAGCATGTACGTAGGTG

Sample: 7840

5' seq:TGGTATTAAGAGAGAAGGAAAAAggtagacttttctaagtggaccaggca
|||||
TGGTATTAAGAGAGAAGGAAAAAATTGATTCAAATTTTCGAGCATGAA
|||||
3' seq:aaagagtataatgtaaaataaaaaATTGATTCAAATTTTCGAGCATGAA

Sample: 8094

5' seq:ATATAGAGACTTTGAATAAGTCGAACActaagaagtacttttgaaggctcg
|||||
ATATAGAGACTTTGAATAAGTCGAACATTTTAGTTTTATAAGACTTAATA
|||||
3' seq:gatgatagaataaaccactacttttagTTTtagTTTTATAAGACTTAATA

Sample: 3973

5' seq:CTTGTCGGGAGCTACCCTGTGTAActgcaggggtgttttgcagcatgcctgg
|||||
CTTGTCGGGAGCTACCCTGTGTAAAGATGTGGCCCCTCAACCTTGGACTTCT
|||||
3' seq:tggatgggagaaggctctgaccaAATGTGGCCCCTCAACCTTGGACTTCT

Sample: 5078

5' seq:CTGAGATTCTATAtttctgacaagctcccagataatgtcaatatgggtgggtccaggacattttgattag
|||||
CTGAGATTCTATActcagattctatactagttctattaatctagttctatactagttctgaGATTCTAAC
|||||
3' seq:caaataatttagtaaaattttctggattatgtagcaagtagttttccttcccaagttcattGATTCTAAC
|||||

Sample: 2707

5' seq:CGGGCCATGTTAAAGAATCCGAAGtataaacacgggttttaacatatatata
|||||
CGGGCCATGTTAAAGAATCCGAACCTTTCTGATGGCACGTAAAACAAATTT
|||||
3' seqcacatcccagattttaaaggttAACCTTTCTGATGGCACGTAAAACAAATTT
|||||

Sample: 4325

5' seq:TGTCTTCCAAGATTCAAATGGCATCCcctaattgcaatgtattatgattt
|||||
TGTCTTCCAAGATTCAAATGGCATCCATATAAAATCTCTATAACTGAGAT
|||||
3' seq:cgaagagtcaggtgagatgatggacgTATAAAATCTCTATAACTGAGAT
|||||

Sample: 1148

5' seq:GGACGGAGTCGGAGGGTTCAAtggaccctaattgtccgtacacgggtgttgcgggtcgattgaaaaaaaaa
|||||
GGACGGAGTCGGAGGGTTCAAttagtgcatcatcatTAGTGCATCATTATTTCGGAATATATAGGTATAA
|||||
3' seq:atcctacgtattttgacggctaagaacctagggttaagTAGTGCATCATTATTTCGGAATATATAGGTATAA
|||||

Sample: 3308

TGTTTTTAACTGTTAGGATACTTGAATccaaagggtaaagaaactcagctgatttatacgtttcttc
|||||
TGTTTTTAACTGTTAGGATACTTGAATTCACAGTTTTTATCTTTGATTGAAATTAATTTAGATT
|||||
agattcaatatgtctaaatgtatataatTCACAGTTTTTATCTTTGATTGAAATTAATTTAGATT

Sample: 5904

TTTTTTCCTCTTTTAAACTATGAAAATACTATTtaaactaatttccaagccatttttaaattaaaaatgtaa
|||||
TTTTTTCCTCTTTTAAACTATGAAAATACTATAatactatTTAGGTATTTGTCCTAATGCTCTCCCTCACCTTT
|||||
aaccgctcatctagggtttatctagggtttaagccccacatgcaTTAGGTATTTGTCCTAATGCTCTCCCTCACCTTT

Sample: 6194

AAAGTGAATTTTAGTTAAGTcatcttattcactttaccgaggaaa
|||||
AAAGTGAATTTTAGTTAAGTttaagtgtttagtttagttaagtgtttagttaagtgtTAACCCAGCTGTGTGTTGG
|||||
attaggcatttccttcccctcaccactcacctctgaTAACCCAGCTGTGTGTTGG

Sample: 0845

TGTGAAAACAACAACAAATTTTGATTGAGACCCTGTatttttcagacctttggtcttgtgaggaaggaa
|||||
TGTGAAAACAACAACAAATTTTGATTGAGACCCTGTACCCTAGCCTTTCCTGCAGCCTTCACTCCCCAA
|||||
agatgggttttctgcatcccagaaggcaatACCCTAGCCTTTCCTGCAGCCTTCACTCCCCAA

Sample: 6730

GGATCCAGTTTCAGCTTTCTACGTATGGCTAGCCAATTttccagcaccggtttattaaatagggaatcct
|||||
GGATCCAGTTTCAGCTTTCTACGTATGGCTAGCCAATTTGACCACAATGATAAGCATACTTATTAGCAT
|||||
ttccacactaaaaatctccagtgaattgggcagTTGACCACAATGATAAGCATACTTATTAGCATGCCTC

Sample: 7573

```
TCTTTTCCATGCCTTTTCCAGCTTTTAGAGCCTGCCTGCATTtcttggctcttggccctgtccttcatctt
|||||
TCTTTTCCATGCCTTTTCCAGCTTTTAGAGCCTGCCTGCATTGACAACAATTAGTGACTIONATAATTAAA
|||||
cttttaatctctgaactaagaaattaaaattgtatTTGACAACAATTAGTGACTIONATAATTAAA
```

Sample: 9723

```
CATGGAATGATAAGGAGGAAGGGAATATCTAACAGCActtttttagatatctctcatagaattgttaatgtgagc
|||||
CATGGAATGATAAGGAGGAAGGGAATATCTAACAGCAAGGGAATCTAGGCAGCATATTTACAGCTTTTCACTAC
|||||
CtcttagcagctggggaactaagtccttcatttccaaAGGGAATCTAGGCAGCATATTTACAGCTTTTCACTAC
```

Sample: 0340

```
TACAAATAACCAAATGTATAAGATGACACCCGAAATTGgcttcaaagccatgcaacctgtgcagttatgcagtaa
|||||
TACAAATAACCAAATGTATAAGATGACACCCGAAATTGTTATGTATGTAATGACAAGTGAAAATACATTATTGT
|||||
ctgcatagccactaaatgtattttaactttaagggggagTTATGTATGTAATGACAAGTGAAAATACATTATTGT
```

Sample: 4210

```
GTTTATTTCACTTAGCATAATGCCTCTAAGATtcacccatattagctcatcttttgatttgctgaattct
|||||
GTTTATTTCACTTAGCATAATGCCTCTAAGATatggAGTTAGGCTAACTCCATATCTACTTGACAAGTAACC
|||||
tataatgagaagggtaatctcagcataaatttcaaaAGTTAGGCTAACTCCATATCTACTTGACAAGTAACC
```

Sample: 4297

```
GTATGTCTGTGGTAGTAGTTCTCAAAGCATAGGCCAGCcatcagcataacctaggaacttttcagagatgcaagtctt
|||||
GTATGTCTGTGGTAGTAGTTCTCAAAGCATAGGCCAGCAATGGCCTCACATCTTTATATGATTTCAGATAAGGTGA
|||||
aggtcagagaactggtttgagcaaacttttatctcgcAATGGCCTCACATCTTTATATGATTTCAGATAAGGTGA
```

Sample: 4528

```
TAGTGATGATAAAATCAATTTGGCATGTTTCACGGACAggtgtgcattttagaaagggtaaagacttggaacgggat
|||||
TAGTGATGATAAAATCAATTTGGCATGTTTCACGGACAtaTTTCCATATTTGGGAAAAAATTCTCTTGACACCAA
|||||
gaccattttccccaatcaatacctatatattcatactcaagtTTTCCATATTTGGGAAAAAATTCTCTTGACACCAA
```

Sample: 6409

```
ATGTGGATTGTTGTTTTGTTTTGTTTTTTTAACCTGACCGTtgccttggctatatgtttgttggtggctagaaaaa
|||||
ATGTGGATTGTTGTTTTGTTTTGTTTTTTTAACCTGACCGTTTGCCATTTTCAGCACAGCTAATAATTTTAGTAGCA
|||||
gttaatattatcacagcagttaatatcactatagcagttagTTGCCATTTTCAGCACAGCTAATAATTTTAGTAGCA
```

Sample: 9250

```
ACATTTTAATTGAAaataaaaagctgcttttattcaaaggaagcaccactgac
|||||
ACATTTTAATTGAAtggtgttgcnntgaacatggcattgcaggtgaattgcaggtgaattgCTTGTGAATGGTGTT
|||||
CctcatccatcagtggaactgaggatgtttccacatcttggtCTTGTGAATGGTGTT
```

Sample: 1094

```
AATAAAAATCAAACAGTGTTCTTAGCATAATTTACGCGTgttggatgaactcctattgaacatatacaatctatata
|||||
AATAAAAATCAAACAGTGTTCTTAGCATAATTTACGCGTGAAACTGGAACCTCCAATGTATTTTCTTATAAACAT
|||||
aataaataaagtggaatgaattatatatgttgctggaagtcAAAACTGGAACCTCCAATGTATTTTCTTATAAACAT
```

Sample: 1314

```
GTGCTACAAAGTGAAGATACATATGGATGGTGAACAACCATgtgttcaccttaacaatgctatattaatggcttgg
|||||
GTGCTACAAAGTGAAGATACATATGGATGGTGAACAACCATTTTACTGATTCTTGGGGAGATTTATCGAGCTACTCA
|||||
gatatccagggtcataagatgttttctccagtggtgccatTTTACTGATTCTTGGGGAGATTTATCGAGCTACTCA
```

Sample: 0020

```
CAGACTTAATGGCTTAAAACAACCCAAATTTATTATCTTcaatTTTggaggtccaaagtccaaaaaatgggtctca
|||||
CAGACTTAATGGCTTAAAACAACCCAAATTTATTATCTTtTAATTAACnGGGGCAAGATTCCTCTGAAATGAGAGA
|||||
gtaaagatatgcttTgtgtttgtgtgcattcaagagacagaaTAATTAACcGGGGCAAGATTCCTCTGAAATGAGAGA
```

Sample: 1154

```
TCTTGGAAGAAAGTAAACATCACATCTGAAAAATAGctgggcattttttcattatctctgcaaggggaggg
|||||
TCTTGGAAGAAAGTAAACATCACATCTGAAAAATAGTAGAGCTTCTCTGAAAAGTGTGTTTACCAAAaCATACTGG
|||||
gacactttgcagcaaggttttaggaaaagctaacgtaagTAGAGCTTCTCTGAAAAGTGTGTTTACCAAAaCATACTGG
```

Sample: 2500

```
TTGGGTTTAATTCCAACCACATACATATAAAACCATGTcagtttcaaagtctaggtcagggctcttaaagctgaaac
|||||
TTGGGTTTAATTCCAACCACATACATATAAAACCATGTCTCTCCTTTACTTTTACCTGCTGCCTTCTCCAGCCT
|||||
aaagtgttgggattacaggcgtgagccaccgtgcctggCTCTCCTTTACTTTTACCTGCTGCCTTCTCCAGCCT
```

Sample: 8094

```
CTATGCTGTACAGTATATCTCTGAAACTTATTCAGCTTGTgattcttcatgaaaacttccagctaatacacttaaa
|||||
CTATGCTGTACAGTATATCTCTGAAACTTATTCAGCTTGTAAAATCAAAATATTCTGAATTATCTTTGGCTTCCTAT
|||||
tttcttcatagtactactatcttattttggtgatgaaaatcAAAATCAAAATATTCTGAATTATCTTTGGCTTCCTAT
```

Sample: 9061

```
AGTGTTAGCTAAGAcATGATACGTATTTCTAGGAGGGAatTTTcaaggcaaggtggagattgtgcagtgacgcca
|||||
AGTGTTAGCTAAGAgATGATACGTATTTCTAGGAGGGAATCTATCACAAGCTGCTTTTCAAAGGCTGCAAGGTGTAGG
|||||
tccttacactcatggatcaagatgactatcagctctctaTCTATCACAAGCTGCTTTTCAAAGGCTGCAAGGTGTAGG
```

Sample: 2326

```
CACCAAGCCCGGCTAATTTGTTTTGTATTTTAGTAGAGatgggggttcaccatggttagccaggatgggtctcgat
|||||
CACCAAGCCCGGCTAATTTGTTTTGTATTTTAGTAGAGACGGGGTTTCACcGTGTTAnCCAGGATGGTCTCaAT
|||||
tgccaccatgcccagctaattttttgtatttttagtagagACGGGGTTTCACtGTGTTAgCCAGGATGGTCTCgAT
```

Sample: 0086

```
CTGCACAATTCTTTTGTCTCATCAAGAGAGCTAGTCagcagacagtaacaaaatgcttatattcatataacctga
|||||
CTGCACAATTCTTTTGTCTCATCAAGAGAGCTAGTCtctcttGCATGGAATGTTCTTCCATTGTTGTGTCCT
|||||
gggcagtatggccattttcacgatattgattcttctatccatgaGCATGGAATGTTCTTCCATTGTTGTGTCCT
```

Sample: 2683

```
TGGGATTaCTCGGCAGTCATCAgAGTTAGATTTAACGActttggagacttaagcattatgggttttttttttttttg
|||||
TGGGATTtCTCGGCAGTCATCAnAGTTAGATTTAACGACTGCCTCCCAACTGAAACACTTCTCTATGAAATCACCAT
|||||
ccatctttcttttacattttaagtcagtttctacatCTGCCTCCCAACTGAAACACTTCTCTATGAAATCACCAT
```

Sample: 6224

```
CCAAGATGGTGAAACCCTGTCTGTACTAAAAATACAAaattagccaggcgcagtggcaggcacctgtaatcccagc
|||||
CCAAGATGGTGAAACCCTGTCTGTACTAAAAATACAAAAACAAACAAAAAAGATTGACTGGATTGTTCTGT
|||||
ctccagcctgggcgactgagcaagactccgtctcaaacAAACAAACAAAAAAGATTGACTGGATTGTTCTGT
```

Sample: 0837

```
GTCCACATTGATGTTGGTTATATAAAATATTCATGctaaatcatataaatgctaaacatttgtatttgtgatg
|||||
GTCCACATTGATGTTGGTTATATAAAATATTCATGATATTAGATATATTTTCATGGATACCATnTATTATAAGCT
|||||
agctatatcaaatgatcttctctccaaattaaaatggATATTAGATATATTTTCATGGATACCATaTATTATAAGCT
```

Sample: 5366

```
ACTGCTGAAAGAAATCATAGATGACACAAACAAATGAaaacacatcccatagtcgtggatgggtacaatcagtat
|||||
ACTGCTGAAAGAAATCATAGATGACACAAACAAATGAtttcactGGTGTGAAATCCTAGCAATGAAAGATGGCAGAG
|||||
caagttgcagagtgattccaaggggtgctcactttaagaagcacgGGTGTGAAATCCTAGCAATGAAAGATGGCAGAG
```

Sample: 5024

```
TTTTCTTTTAATGTTTAATCGACACATAATTATGCATATttaggaggtacagagtgatattttcctatatgaatataacatgtaataatcaaatc
|||||
TTTTCTTTTAATGTTTAATCGACACATAATTATGCATATattatgcatatataaatatgcataattatgcatatttatATCTCTCCAGATATAAAA
|||||
ttattccttgcaagtagcaagtggttaagagcaaagctacaaaatacaagttttataATCTCTCCAGATATAAAA
```

Sample: 1505

```
GGGCTTACTACTGAGATTGTGAAGATATATATATATATctgtcagctgtgtctcagcaggggaacaaagttgagatat
|||||
GGGCTTACTACTGAGATTGTGAAGATATATATATATATatCATTTTTGTnGGGCCATTTTTGTTCTGTTTCCnGGn
|||||
aactgcaattttcttttgcaccaacctaatatatctcagtcCATTTTTGTaGGGCCATTTTTGTTCTGTTTCCaGGt
```

Supplementary Data S3. Breakpoint junction sequence alignment and microhomology determination. For each case, both 5' and 3' reference human sequence are aligned with the patient sequence across the breakpoints as sequenced. The Red boxes represent the microhomologies observed at the deletion breakpoint junctions.