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Supplemental Data

***BMPER* Mutation in Diaphanospondylodysostosis**

Identified by Ancestral Autozygosity Mapping

and Targeted High-Throughput Sequencing

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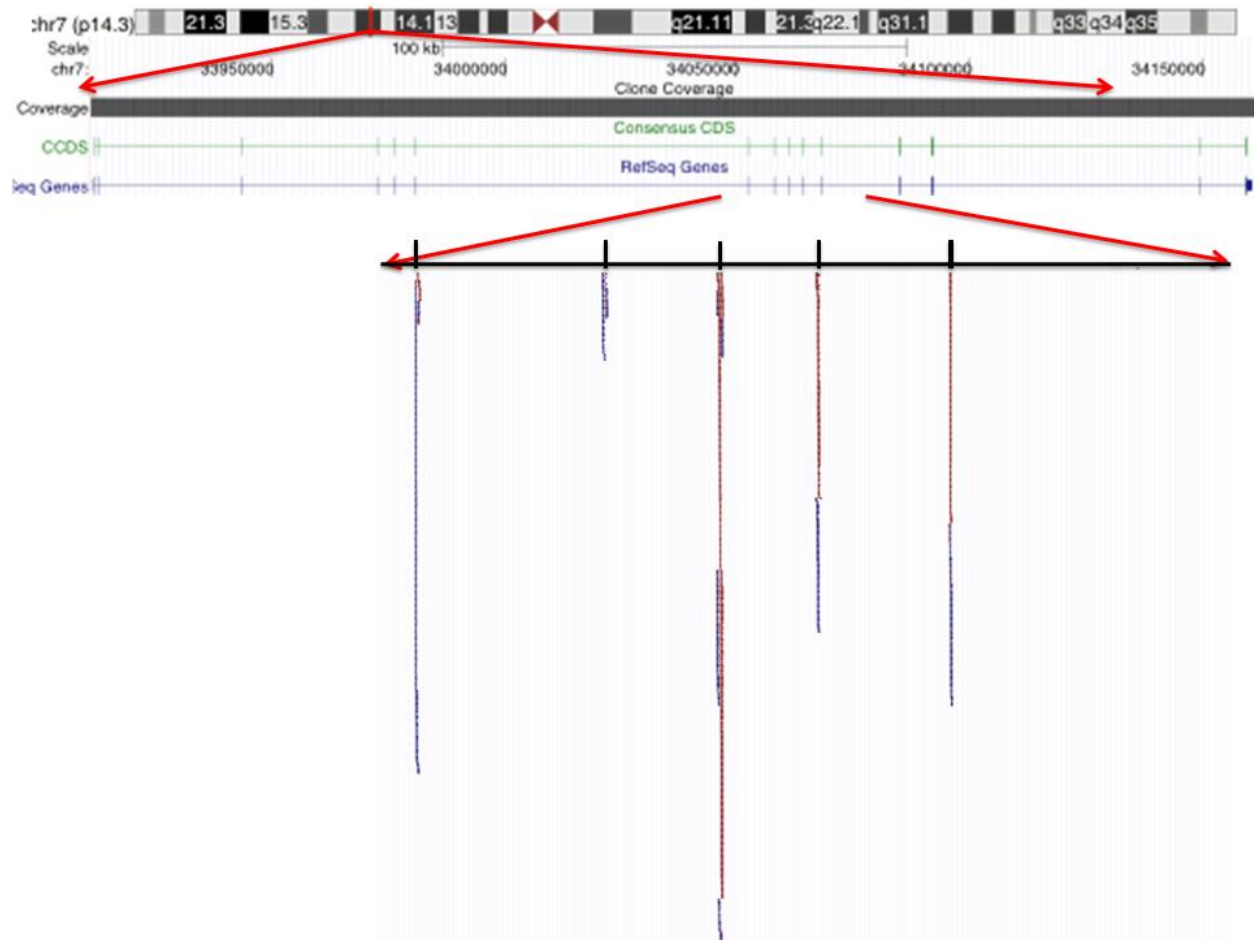


Figure S1. Specificity of Array Based Custom Target Enrichment

At the top, the genomic structure and location of *BMPER* on chromosome 7 is shown. Below, a zoomed view of 25 kb of *BMPER* shows the specificity of capture and sequencing for exons 7-11. Note that very few sequences were derived from genomic regions outside of the targeted exons. Forward and reverse sequences of fragments derived from the Solexa sequences are shown in red and blue, respectively, for the final 2 codons and first 3 bp of exon 9 and the succeeding intron 9. Homozygosity for the pathological sequence change c.925C>T which results in p.Q309X is shown. Confirmation of the mutation derived from sequence analysis of a PCR amplified fragment containing exon 9 from R05-062A is shown at the bottom.

Table S1. Runs of Homozygosity (ROH) in Affected and Unaffected Siblings

Chr ^a	Affected ROH (Mb) (R05-062A)	Unaffected Sib (ROH) (R05-062D)	ROH >3MB (MB)	ROH in Affected Only	Total Genes
1		7.414-9.250			
1	18.424-20.767				
1 ^b	163.458-176.256		13	13	108
2		124.610-127.614			
3		63.366-68.863			
4		141.007- 154.354			
4		171.033-174.323			
5		2.705 -9.738			
5	31.353-32.296				
5		157.520-160.470			
6	112.409-124.223	109.290-127.977	12		N/A
7 ^b	33.778-55.173		21	21	105
9	107.099-108.283				
9	128.782-129.883				
12		3.332-4.825			
12		91.033-93.087			
17 ^b	32.306-36.011		4	4	69
17		44.865-59.744			
18		10.585-13.236			
18	54.396-56.338	54.471- 55.673			
TOTAL	57.226	59.235	50	38	282

^a Chromosome where run of homozygosity was detected in affected and unaffected siblings.

^b Runs of Homozygosity(ROH) which were significantly long, considered autozygous in affected sibling and not also identified in the unaffected sibling.

Table S2. Summary of All Variants Identified by Targeted High-Throughput Sequence Analysis in Diaphanospondylodysostosis Case II-2

Number of variants detected	1,915
Known SNPs (dbSNP130)	1,749
Unknown variants in exons	13
Unknown stop	1

Table S3. Primer Sequences for Amplification of the *BMPER* Exons

Exon	Primer Name	Sequence	Product Size (bp)	hg18 Chromosome Location (bp)
1	BMPER 1F	gagagccctttcgactgtgag	438	chr7:33911663-33911684
	BMPER 1R	ttattactacattgaaggcaacagc		chr7:33912076-33912100
2	BMPER 2F	aattctgaagtttggtagaggttt	300	chr7:33912842-33912866
	BMPER 2R	gataaagacaatttgaagatctgg		chr7:33913117-33913141
3	BMPER 3F	acctctgtggtgtgtaggatttg	250	chr7:33943350-33943373
	BMPER 3R	ggaaggtgagagaaagagaaataa		chr7:33943576-33943599
4	BMPER 4F	cttattttgctccaaaaggtaaagg	249	chr7:33972539-33972563
	BMPER 4R	tagggttttgttctatgtgtgtga		chr7:33972763-33972787
5	BMPER 5F	ctcctacttaggtcactttccaaaa	299	chr7:33976385-33976409
	BMPER 5R	gctctctaaaccattaggtacaca		chr7:33976659-33976683
6	BMPER 6F	gatgaactgtggatagaaggattgt	264	chr7:33980757-33980781
	BMPER 6R	gagacgagagagaggctgtagaga		chr7:33980997-33981020
7	BMPER 7F	taatactatggaagggccttaggtt	285	chr7:34052341-34052365
	BMPER 7R	aaaacaacaacgtggataaacactc		chr7:34052601-34052625
8	BMPER 8F	ataatggacctatgatggaatcacc	297	chr7:34057911-34057935
	BMPER 8R	caataaataacacaccattggcttt		chr7:34058183-34058207
9	BMPER 9F	agttatgaaaggaaggccaggtag	297	chr7:34061219-34061242
	BMPER 9R	ctgcctagtgtctggagcttttta		chr7:34061492-34061515
10	BMPER 10F	ccatggagaggttacagtctgttg	281	chr7:34064117-34064140
	BMPER 10R	agctggagtctgtgttcggaagat		chr7:34064374-34064397
11	BMPER 11F	ttggactgatttcattggtttatct	248	chr7:34068021-34068045
	BMPER 11R	ggaaacatacaccctatttttctt		chr7:34068244-34068268
12	BMPER 12F	agttgatttcccttagtgcatttct	542	chr7:34084874-34084898
	BMPER 12R	accacatctcctgagtgtcttaac		chr7:34085392-34085415
13	BMPER 13F	tcatatgttctgtgtaaggatgat	500	chr7:34091804-34091828
	BMPER 13R	catacaccatgttccaaggtaag		chr7:34092280-34092303
14	BMPER 14F	cgacccttctgaagttatatttagg	300	chr7:34149286-34149312
	BMPER 14R	actcatgtcagcatccacaagtct		chr7:34149562-34149586
15	BMPER 15F	ttaaagtcaaatgtgatcctgag	400	chr7:34159094-34159118
	BMPER 15R	cacaaatactgcagtccttcattg		chr7:34159470-34159494