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# Supplementary Information for

The Landscape of submicroscopic structural variants at the OPN1LW/OPN1MW gene cluster on Xq28 underlying Blue Cone Monochromacy: Evidence for the instability of gene clusters with increased copy number

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## Other supplementary materials for this manuscript include the following:

Dataset S1

## Fig. S1. (below). Breakpoint sequences of SVs.

Sanger sequencing electropherograms were obtained from PCR amplicons covering the SV breakpoints. Breakpoint junctions are indicated by stippled vertical lines and the physical coordinates of the last nucleotide upstream and the first nucleotide downstream of the junction are indicated. The mutant allele sequence (SV-seq) is given below and aligned with the centromeric and telomeric reference sequences flanking the breakpoint. Matching reference sequence upstream and downstream of the breakpoint in bold. Sequence homologies at the breakpoint junctions are highlighted in yellow. Inserted sequences at the breakpoint junctions are indicated by red letters and additional deletions by dashes. Note that the breakpoint sequences of SVar1, SVar20, SVar38, and SVar42 are shown in Figures S2-S4.































































**Fig. S2. Complex structure of SVar42.** (A) Genomic structure of SVar42 in comparison with the normal chromosomal structure (top). SVar42 constitute a deletion of the entire *OPN1LW/MW* gene cluster downstream of the intact LCR. The deletion further includes the downstream *TEX28* and *TKTL1* genes and is combined with an interstitial inverted duplication of about 80 kB including eight annotated genes from *RPL10* to *LAGE3* (Dup-Inv). Deleted sequences are depicted in light shading and stippled lines flanked by brackets. The inverted and duplicated segment is inserted upstream of *FLNA* and *EMD* and their flanking low copy inverted repeats (LR<sub>A</sub> and LR<sub>B</sub>, black horizontal arrows). The telomeric repeat LR<sub>A</sub> is thereby converted into a LR<sub>B</sub>-like repeat copy (LR<sub>B'</sub>). (B) Sanger sequence electropherogram of the centromeric deletion breakpoint adjunct to the 394bp insertion (derived from multiple small remnant sequences, light grey box) and the terminus of the inverted duplication (rose box).

**Fig. S3. (below).** Insertions at breakpoints are short remnants of OPN1LW/MW gene cluster sequences suggestive for a replication-based mechanism of SV formation. Results on the origin of breakpoint inserted sequences for SVar3, SVar24, SVar28, SVar36, and SVar42. For each SV the inserted sequence is provided with parts showing perfect or near perfect match with sequences of the *OPN1LW/MW* gene cluster and flanking sequences highlighted with colored boxes. The location (e.g. upstream *OPN1LW*), orientation on the sense (+) or complementary strand (-), and physical coordinates of the matching genomic sequences are provided. The 'Sbjct' sequence for the alignment is always NC\_000023.11.

## SVar3

NC\_000023.11: g.154139421\_154142962delins19

#### Inserted Sequence: CCTGGAGGTGCACATGCAG

#### Upstream OPN1LW(-)

Query	1	GGTGCACATGCAG	13
Sbjct	154143629	GGTGCACATGCAG	154143617

#### SVar24

NC\_000023.11:g.154136800\_154180989delins51

## Inserted Sequence: TGCTAAGGCAGACCAGAAGGCGCCCACTGCGCCTGTTGACAGGTGGGCGCC

#### Upstream OPN1LW(+)

Query	1	TGCTAAGGCAGACCAGAAGGCGCCCAC	27
Sbjct	154136924	TGCTAAGGCAGAC.AGAAGGCGCCCAC	154136949

#### SVar28

NC\_000023.11:g.154139374\_154206273delins139

#### Inserted Sequence:

TGGGAAACAAGAAATCACAATACGGTGCTCTGTGAGCCGCAAAGTGTGGGAGAATTGGACGTGCAT<mark>TCTCTCC</mark> CAATTGGCCACTGTTCCGGTTTCTGTCACT</mark>GGGC<mark>GTGTGTCAGGGGGGGCCACAGTGAGGAGGCAG</mark>G

#### Intergenic(+)

Query	1	TGGGAAACAAGAAATCACAATACGGTGCTCTG	TGAGCCGCAAAGTGTGGG	50
Sbjct	154176356	TGGGAAACAAGAAATCACAATACGGTGCTCTG	TGAGCCGCAAAGTGTGGG	154176405
Sbjct	154213494	TGGGAAACAAGAAATCACAATACGGTGCTCTG	TGAGCCGCAAAGTGTGGG	154213543
Sbjct	154251296	TGGGAAACAAGAAATCACAATACGGTGCTCTG	TGAGCCGCAAAGTGTGGG	154251345
Sbjct	154288414	TGGGAAACAAGAAATCACAATACGGTGCTCTG	TGAGCCGCAAAGTGTGGG	154288463
Query	51	AGAATTGGAC 60		
Sbjct	154176406	AGAATTGGAC 154176415		
Sbjct	154213544	AGAATTGGAC 154213553		
Sbjct	154251346	AGAATTGGAC 154251355		
Sbjct	154288464	AGAATTGGAC 154288473		
Interge	enic(-)			
Query	69	TCTCCCAATTGGCCACTGTTCCGGTTTCTG	TCACT 103	
Sbjct	154168994	TCTCCCATAATTGGCCACTGTTCCGGTTTCTG	TCACT 154168958	
Sbjct	154206122	TCTCCCATAATTGGCCACTGTTCCGGTTTCTG	TCACT 154206086	
Sbjct	154243931	TCTCCCATAATTGGCCACTGTTCCGGTTTCTG	TCACT 154243895	
Sbjct	154281052	TCTCCCATAATTGGCCACTGTTCCGGTTTCTG	TCACT 154281016	
Interge	enic(-)			
Query	108	GTGTGTCAGGGGAGCCACAGTGAGGAGGCAG	138	
Sbjct	154173338	GTGTGTCAGGGGAGCCACAGTGAGGAGGCAG	154173308	
Sbjct	154210476	GTGTGTCAGGGGAGCCACAGTGAGGAGGCAG	154210446	
Sbjct	154248278	GTGTGTCAGGGGAGCCACAGTGAGGAGGCAG	154248248	
Sbjct	154285396	GTGTGTCAGGGGAGCCACAGTGAGGAGGCAG	154285366	

#### SVar36

#### NC\_000023.11:154120448\_154281284delins180

#### **Inserted Sequence:**

# Intergenic(-)

7	CCCACCCCTTAACACCATCACATGGCCATTAAATTTCAACACGAGTTTTG	56
154168440	CCCACCCCTTAACACCATCACATGGCCATTAAATTTCAACACGAGTTTTG	154168391
154205568	CCCACCCCTTAACACCATCACATGGCCATTAAATTTCAACACGAGTTTTG	154205519
154243377	CCCACCCCTTAACACCATCACATGGCCATTAAATTTCAACACGAGTTTTG	154243328
154280498	CCCACCCCTTAACACCATCACATGGCCATTAAATTTCAACACGAGTTTTG	154280449
	7 154168440 154205568 154243377 154280498	7CCCACCCCTTAACACCATCGCCATTAAATTTCAACACGAGTTTTG 

Query	57	GGGGGGGACATGTACCCCATAGCAGTATGCTTAACTTTTTAAGAAAGA	106
Sbjct	154168390	GGGGGGACATGTACCCCATAGCAGTATGCTTAACTTTTTAAGAAAGA	154168341
Sbjct	154205518	GGGGGGACATGTACCCCATAGCAGTATGCTTAACTTTTTAAGAAAGA	154205469
Sbjct	154243327	GGGGGGACATGTACACCATAGCAGTATGCTTAACTTTTTAAGAAAGA	154243278
Sbjct	154280448	GGGGGGACATGTACCCCATAGCAGTATGCTTAACTTTTTAAGAAAGA	154280399
Query	107	AGGCCGGGCGCGGTGGCTCACCCCTGTAATCCCAGCACTTTGGGAGGCTG	156
Sbjct	154168340	AGGCCGGGCGCGGTGGCTCACCCCTGTAATCCCAGCACTTTGGGAGGCTG	154168291
Sbjct	154205468	AGGCCGGGCGCGGTGGCTCACCCCTGTAATCCCAGCACTTTGGGAGGCTG	154205419
Sbjct	154243277	AGGCCGGGCGCGGTGGCTCACCCCTGTAATCCCAGCACTTTGGGAGGCTG	154243228
Sbjct	154280398	AGGCCGGGCGCGGTGGCTCACCCTGTAATCCCAGCACTTTGGGAGGCTG	154280349
Query	157	AGGCAGGCGGATCACCTGAGGTCG 180	
Sbjct	154168290	AGGCAGGCGGATCACCTGAGGTCG 154168267	
Sbjct	154205418	AGGCAGGCGGATCACCTGAGGTCG 154205395	
Sbjct	154243227	AGGCAGGCGGATCACCTGAGGTCG 154243204	
Sbjct	154280348	AGGCAGGCGGATCACCTGAGGTCG 154280325	

#### SVar42

NC\_000023.11:g.154143928\_154338057delins[394;g.154394091\_154480236inv]

#### **Inserted Sequence:**

TGAACTGGCTCATCCACCAGAACGCCAAAAATTAAAAAG<u>CCT</u>GCCCCAAAGGCGGACGCAGGACAGTA GAAGGGAACAGAGAACACATAAACACAG</mark>CCATAGACCTGG<mark>GCCTGGGCCCCGACTGGCTTACCACACA GG</mark>CAGCCCCTCCCT<mark>TGGGTGTTGGGAACCAAACTCAGACGCCCCACCCATCCCCGCCAG</mark>TGGCTTGAT CTCAGGAGCAGCTGGGCCCAGTCGCTAAAAGTATGCAGCTGGATCCTGGCAGAGACCG<mark>TCATTCACCC TGCAAGCCCCTCCGGCC</mark>TGGGCAGCAGAGCAAGACACCA<u>CC</u>AGGACACATAGGGTGGCCCAGCACG AAGTAGCCAG

#### Intergenic(+)

9	CTCATCCACCAGAACGCCAAAAATTAAAAAGCCT	42
154181862	CTCATCCACCAGAACGCCAAAAATTAAAAAGCCT	154181895
154219001	CTCATCCACCAGAACGCCAAAAATTAAAAAGCCT	154219034
154256805	CTCATCCACCAGAACGCCAAAAATTAAAAAGCCT	154256838
154293923	CTCATCCACCAGAACGCCAAAAATTAAAAAGCCT	154293956
	9 154181862 154219001 154256805 154293923	9         CTCATCCACCAGAACGCCAAAAATTAAAAAGCCT           1

#### Upstream OPN1LW(+)

Query	40	CCTGCCC	AAAGGCGGACGCAGGACAGTAGAAGGGAACAGAGAACACATA	89
Sbjct	154143923	CCTGCCCC	AAAGGCGGACGCAGGACAGTAGAAGGGAACAGAGAACACATA	154143972
Query	90	AACACAG	96	
Sbjct	154143973	AACACAG	154143979	

## OPN1LW/MW exon 2 & intron 2(-)

0	100		1 2 0
Query	109	GCCTGGGCCCCGACTGGCTTACCACACAGG	138
Sbjct	154150974	GCCTGGGCCCCGACTGGCTTACCACACAGG	154150945
Sbjct	154188088	GCCTGGGCCCCGACTGGCTTACCACACAGG	154188059
Sbjct	154225226	GCCTGGGCCCCGACTGGCTTACCACACAGG	154225197
Sbjct	154263030	GCCTGGGCCCCGACTGGCTTACCACACAGG	154263001

## Upstream OPN1LW(-)

Query	158	TGGGAACCAAACTCAGACGCCCCACCCATCCCCGCCAG	195
Sbjct	154144061	TGGGAACCAAACTCAGACGCCCCACCCATCCCCGCCAG	154144018

## OPN1MW intron 2(+)

Query	263	TCATTCACCCTGCAAGCCCCTCCGGCC	289
Sbjct	154188103	TCATTCACCCTGCAAGCCCCTCCGGCC	154188129
Sbjct	154225241	TCATTCACCCTGCAAGCCCCTCCGGCC	154225267
Sbjct	154263045	TCATTCACCCTGCAAGCCCCTCCGGCC	154263071
Sbjct Sbjct Sbjct	154188103 154225241 154263045	IIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIII	1541881 1542252 1542630

## Intergenic(+)

287	GCC <mark>TGGGCAGCAGAGCAAGACACCACC</mark>	313
154165572	GCCTGGGCAGCAGAGCAAGACACCACC	154165598
154202686	GCCTGGGCAGCAGAGCAAGACACCACC	154202712
154240544	GCCTGGGCAGCAGAGCAAGACACCACC	154240570
154277630	GCCTGGGCAGCAGAGCAAGACACCACC	154277656
	287 154165572 154202686 154240544 154277630	287GCCTGGGCAGCAGAGCAAGACACCACC154165572GCCTGGGCAGCAGAGCAAGACACCACC154202686GCCTGGGCAGCAGAGCAAGACACCACC154240544GCCTGGGCAGCAGAGCAAGACACCACC154277630GCCTGGGCAGCAGAGCAAGACACCACC

## OPN1LW exon 2(-)

Query	312	CCAGGACACACATAGGGTGGCCCAGCACGAAGTAGCCAG	350
Sbjct	154150934	CCAGGACACACATAGGGTGGCCCAGCACGAAGTAGCCAG	154150890

## Upstream OPN1LW(+)

Query	347	CCAGCAAATCCCTCTGAGCCGCCCTTGCGGGCTCGCCTCAGGAGCAGG	394
Sbjct	154144053	CCAGCAAATCCCTCTGAGCCGCCCTTGCGGGCTCGCCTCAGGAGCAGG	154144100



Fig. S4. Identification of a concurrent 142bp deletion (SVar1) in exon 5 of OPN1LW and **OPN1MW** in family BCM 262. (A) Pedigree of family BCM 262 showing the affected index patient (III:2) by a black square. There is a history of colorblindness with preserved vision for the deceased maternal grandfather. (B) Genotyping strategy of the OPN1LW and OPN1MW exon 5 on the normal X chromosome  $(X_N)$  and the BCM-linked X chromosome  $(X_{BCM})$  by either non-discriminating one-step amplification of exon 5 (black line) or using long distance PCR for separate amplification of OPN1LW (LD-Proximal, red line) and OPN1MW (LD-Distal, green line) gene fragments, followed by a nested PCR of exon 5. (C)-(E) Agarose gels showing PCR products for subjects I:1, II:2, III:1, and III:2 obtained by non-discriminating direct amplification of exon 5 (C), and nested amplification after long distance PCR of OPN1LW (D) or OPN1MW (E). Sanger sequencing traces of the respective amplicons from III:2 are mounted below. (F) Alignment of parts of the OPN1LW and OPN1MW cDNA sequences (differences highlighted with red and green letters) along with the sequence of the SVar1 deletion (deleted nucleotides indicated by dashes). LCR: Locus Control Region, LD: Long-Distance, Re-Exon 5: re-amplification of exon 5; M: Marker ladder, Ex5: Exon 5, Del: Deletion, NC: Negative Control.

Α

>hg38\_dna range=chrX:154136242-154137008 5'pad=0 3'pad=0 strand=+
repeatMasking=none

TTGTATGTGC <mark> </mark> AAATCTTGGTATTTTAATTGCATATGTATCGTATAAATTA	<mark>SVar21</mark>
AGCTTTTGTCTGATTTCCACTGTCTGTAAGAGTTAAAATTAAAAAAAA	
ACACTTGGCTGGACCACACACTGAATTAATGATTTTAACACAAAAATGAT	
ACTGAAGAGAATGTGAATGCACTAATGCCACCGAACTGTGTGTACTTAAA	
AACGGTTAAGATGGTAAGTTGTATGTTATGGGTATTTTACCACTATTAAA	
AAAAAGTTTTTAAGAAA AAAGGCTAATAAGATTTCTGGGTTCAGGCAGGA	<mark>SVar22</mark>
GTAGGAAGGCAGCGAAAACCCGAATTCCCCCACTTCTCCTAAAAGAAGTC	
CAAACACAGGAAGAATGAATGAGCCCCCCCTCCTTGATATACTGGAGGAAA	
CCAGAACCTGTGCCATCAGAAAGCGCTAGGATGTGACAAAGCAGGGAGGG	
GACTACAAAGGGCCCCAGCGGTCCCGACCAGGATCCACCCTTTCAGGACA	
TGGCCCAGGCCCGCTCAGGATC GCATGGGGGCAGATGCGAGCCCCAGGAG	<mark>SVar23</mark>
AACTCCCG AGGACCCGCGGCGGCCGCCGGGGGGCTTCCTGACGCGCGAGCG	<mark>SVar24</mark>
GCGAGGCGGCCAGAAGAGGGCGCCCGGGAGCCGAACAGGAGG	
CCCGGGCCTTCCCGCGTTCCCGGGGGGTCCCCGTGCTAAGGCAGACAGA	
GCGCCCACCCTGTCAACAGTGAGAACCAGAATCCACAAAGGCCAAAGTGCG	<mark>SVar25</mark>
GAAGGC ACTGAGACAAA	<mark>SVar39</mark>



Fig. S5. GC content at the 5' breakpoint cluster at chrX:154136242\_154137008.

(A) Six centromeric breakpoints (indicated by vertical yellow lines) cluster within a 750 bp region 7.2-8 kb upstream of *OPN1LW* which shows a steep increase in GC content from below 25% to higher than 75% (B). GC content (Y-axis) calculated by a sliding window of 50bp size.

В

							1		1															-
	ition	BCM 63	BCM 127	BCM 152	BCM 157	BCM 158	BCM 162	BCM 227	BCM 264	BCM 232	BCM 128	BCM 150	BCM 141	BCM 2	BCM 18	BCM 118	BCM 124	BCM 165	BCM 213	BCM 243	BCM 294	BCM 222	BCM 156	
< Marker	< Phys. Pos (Mb)	#15108	#20621	#22676	#22586	#22961	#23330	#27018	#28186	#27291	#20624	#22574	#21876	#6679	#9325	#20544	#20580	#23548	#26379	#27935	#29729	#26657	#22859	
DXS 8011	150.69	8	8	8	8	8	8	8	7	4	8	8	5	3	1	1	1	1	1	1	1	2	7	
DXS 8103	150.94	2	2	2	2	2	2	2	2	3	4	4	1	3	2	2	2	2	2	n.d.	2	2	2	
DXS 1356	153.43	2	2	2	2	2	2	2	1	2	2	2	2	2	2	2	2	2	2	2	2	2	2	
DXS 8087	153.62	1	1	1	1	1	1	1	1	4	1	1	1	1	1	1	1	1	1	1	1	1	1	
DXLD 15535	154.04	2	2	2	2	2	2	2	1	2	2	2	2	2	2	2	2	2	2	2	2	2	2	
DXLD 36169	154.13	3	n.d.	3	3	3	3	3	3	3	3	3	3	3	3	3	3	3	3	n.d.	3	3	3	
L441 TA	154.51	2	2	2	2	2	2	2	2	2	2	2	2	2	2	2	2	2	2	n.d.	2	2	2	
L441 CA	154.52	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	n.d.	1	1	1	
AF277A	154.55	2	2	2	2	2	2	2	2	2	2	2	2	2	2	2	2	2	2	n.d.	2	2	2	
AF277B	154.60	2	2	2	2	2	2	2	2	2	2	2	2	2	2	2	2	2	2	2	2	2	2	
DXS 1073	154.60	2	2	2	2	2	2	2	2	2	2	2	2	2	2	2	2	2	2	2	2	2	2	

## Fig. S6. (previous page). SVar26 is a founder mutation in BCM families from the United

**States**. Eleven microsatellite markers (left column, encompassing a region of about 3.9 Mb on Xq28) were genotyped in the index patients of 22 families sharing the SVar26 deletion. Alleles (coded in numbers) of identical size are depicted as blue squares and deviant haplotype segments shown in different colors. The localization of the *OPN1LW/OPN1MW* gene cluster is indicated by the red arrow on the right. The index patients from all 22 families share a common haplotype in the vicinity of the *OPN1LW/OPN1MW* gene cluster (from DXLD36169 to DXS1073, encompassing a region of 0.47 Mb) with several recombinations telomeric to the gene cluster. n.d. – not determined

										_
	tion	BCM 138	BCM 146	BCM 153		tion	BCM 197	BCM 198	BCM 245	
< Marker	< Phys. Posi (Mb)	#21475	#22040	#22677	< Marker	< Phys. Posi (Mb)	#24863	#24872	#28091	
DXS 8011	150.69	1	8	1	DXS 8011	150.69	6	6	6	
DXS 8103	150.94	3	2	2	DXS 8103	150.94	2	2	2	
DXS 1356	153.43	3	3	3	DXS 1356	153.43	2	3	2	
DXS 8087	153.62	2	2	2	DXS 8087	153.62	1	1	1	
DXLD 15535	154.04	4	4	4	DXLD 15535	154.04	3	3	3	
DXLD 36169	154.13	1	1	1	DXLD 36169	154.13	1	1	1	
L441 TA	154.51	2	2	2	L441 TA	154.51	1	1	1	
L441 CA	154.52	1	1	1	L441 CA	154.52	1	1	1	
AF277A	154.55	1	1	1	AF277A	154.55	2	2	2	
AF277B	154.60	2	2	2	AF277B	154.60	2	2	2	
DXS 1073	154.60	2	2	2	DXS 1073	154.60	2	2	2	

**Fig. S7. SVar19 and SVar28 are founder mutations in BCM families from the US and France, respectively**. Eleven microsatellite markers (left column, encompassing a region of about 3.9 Mb on Xq28) were genotyped in the index patients of three families sharing the SVar19 (left) and three families sharing SVar28 (right). Alleles (coded in numbers) of identical size are depicted as colored squares and deviant alleles are depicted as while squares. The localization of the *OPN1LW/OPN1MW* gene cluster is indicated by the red arrow on the right.



**Fig. S8. Genetic refinement of the Locus Control Region.** SVar2 with a deletion of 411 bp at the locus control region (LCR) is to date the smallest deletion abrogating expression of the *OPN1LW* and *OPN1MW* genes – as concluded from the BCM phenotype of the patient. SVar2 overlaps with the HS102 deletion (Nathans et al. 1989) by only 358 bp thus refining the sequence most likely crucial for enhancer activity. This coincides with the peak of sequence conservation during vertebrate evolution (UCSC genome browser snapshot depicted in the box). This refinement may actually enable shortening of enhancer/promoter sequences for cone photoreceptor transgene expression in gene therapy trials. The extent of currently used enhancer sequences in the HR2.1 and HR1.7 artificial promoters is shown above.

Aberration type	Copy Number State	Chr	Cytoband(s)	Marker boundaries	Size (kbp)	Marker Count	Genes in aberration	OMIM Genes
Loss	0	chrX	q28	154,143,428- 154,332,392	189	148	Count:6 Genes List:OPN1LW,OPN1MW2,OP N1MW,OPN1MW3,TEX28,T KTL1	Count:4 OMIM Genes List:OPN1LW (300822), OPN1MW (300821), TEX28 (300092), TKTL1 (300044)
Gain	2	chrX	q28	154,395,688- 154,483,524	88	172	Count:11 Genes List:RPL10,SNORA70,DNAS E1L1,TAZ,CH17- 340M24.3,ATP6AP1,GDI1,FA M50A,MIR6858,PLXNA3	Count:8 OMIM Genes List:RPL10 (312173), DNASE1L1 (300081), TAZ (300394), ATP6AP1 (300197), GDI1 (300104), FAM50A (300453), PLXNA3 (300022), LAGE3 (300060)

## Table S1. Cytoscan HD probe CGH-array based CNV calls on the X chromosome for proband BCM99/#8066 carrying SVar42

Variant	Forward Primer Sequence (5'-3')	Reverse Primer Sequence (5'-3')	Size (bp)
SVar1	TCCAACCCCCGACTCAACTATC	ACGGTATTTTGATGTGGATCTGCT	173
SVar2	ACGTGCCACATTCACACTCTGT	TGTCCCTGTTCTGTGGATGAGA	1382
SVar3	AGGATGTCAATGGCTGTGAGG	TGTTCTCAGAGCCACGATTCTC	988
SVar4	AAATCTCGCCAGGCACCTTG	CCGTGGCTACCCTTTAATGTCT	865
SVar5	AGATAATAGGGCAATGCTTTCCA	CCTCTGGTGCATCACAAACTG	286
SVar6	TATGATCCGCAAGGAGCCGT	CAGAGCTGGAAGAAGCCAAGAG	957
SVar7	ATAATGTATGATATGGAGCATCGT	CATGAACGACTTGCTTGCTTC	867
SVar8	TCTTCTTTGGGGAGGTTTCTCT	CAGAGTGAGCTCATCCATCCTG	728
SVar9	CTGCTGCTTGCTTTCTTTTGAT	TCCCGCCAGTAATGATTGAG	604
SVar10	TCGACCCAGAATTAACCTCTCT	AGGAGTCTCAGTGGACTCAT	1160
SVar11	TTATAGGTGCACGCCACCAG	(same as SVar7 reverse)	1063
SVar12	ACATGGCAAAACTGGGTCTCT	TCACTTAGTGGCCTGGAGGAT	340
SVar13	ATCACTGAAAGAGGCCGTGA	TGAATTCTTACATTTGTCAGTGGTG	815
SVar14	GAGGATCAGTCAAGCCCAAGA	CCTGGAAGGACTGTGACTGCTT	659
SVar15	(same as SVar14 forward)	AGGTTGATGAGCTCAGGGATT	791
SVar16	AAAAGTGACAATGGGAACACTG	(same as SVar15 reverse)	493
SVar17	CCAAGGCAGGAGTATCATTTG	GACCACTACCCACCTCTGTCA	921
SVar18	AACATGGGAATGGATGGA	(same as SVar6 reverse)	295
SVar19	TCTGTGCAGGCTGGATAGTG	GAAGCCTCCAGAACCCTTCAGT	488
SVar20	(same as SVar19 forward)	TGCCAAAAGGGGAACTTCATAG	1292
SVar21	(same as SVar19 forward)	GTGAATGAGTGGTTTCCGCC	1258
SVar22	ACTTGGCTGGACCACACACT	CCTTCTAAGGCCCCAGTTACA	331
SVar23	CTAGCAATGAAAGACAGTTAAAGGGAATCAT	(same as SVar7 reverse)	773
SVar24	GAGTACAGGTATTTGCCACTAAGC	AGGAGTCTCAGTGGACTCAT	539
SVar25	(same as SVar 22 forward)	TACCAGTCCCACCCTTAACAC	1520
SVar26	GGCTTTGGGGGGATATATCTTTTTGAAG	CCTGCAGTCTAAAAGTCATGGT	1494
SVar27	(same as SVar3 forward)	TTCTCTCAGAATGTGGCAGGAC	930
SVar28	(same as SVar3 forward)	ATGGCAGCCAGAGGAAACTC	1249
SVar29	ACGTGCCACATTCACACTCTGT	(same as SVar20 reverse)	1353
SVar30	CCAAAGGACCTACAGCTCATGG	AAAGGAATTTGGTTTGGAGGAG	417
SVar31	GATTGGGTGGCTTTTAAATGAT	TCAGACGAGGCTTGGAGATAA	688
SVar32	GTAACTCCTATGTGTGACAGAAG	AGGGAGGGAGGGGGTGTTATCT	723
SVar33	ACAAACCCCACCCGAGTTAG	CTTCCCTCTCCTCACTTTTCCATTC	1332
SVar34	TCACAGCAGGAAAACAGACCTA	CTACTTGCCTCCTGCTTCCC	2280
SVar35	GCAGATGCCCTAGAGTTTGC	(same as SVar5 reverse)	870
SVar36	CCTAGAGTTTGGTTTGAACGGA	GTTTTGCTCTTGTTGCCCAGA	455
SVar37	CTAGAAGAGGCCTGTCATCCCAG	GGCCATTGCCTTGTATTTTT	780
SVar38	TCTGTGCAGGCTGGATAGTG	(same as SVar20 reverse)	1292
SVar39	CGCTAGGATGTGACAAAGCAG	(same as SVar25 reverse)	990
SVar40	GGCATAGAGTTGTTCATAGGATTTC	ACTCTTCCAGATCAGGGATTGA	480
SVar41	GCAGCCTGGACAACATAACA	СТССССАСААСТСССТАТ	1148
SVar42	CTGGGCTTTCAAGAGAACC	GCCTGGTTGATAGAGCCAGACC	588

# Table S2. PCR primers for diagnostic PCRs