

Supplemental information

**Whole-exome sequencing improves
the diagnosis and care of men
with non-obstructive azoospermia**

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Supplementary table 1: Origin and clinical information on all included patients.

sampleID	Country of origin	Testicular Phenotype	TESE
P0074	Algeria	MeA	NEG
P0075	Tunisia	SCOS	NEG
P0080	Algeria	HS	POS
P0081	Algeria	SCOS	NEG
P0082	Algeria	MeA	NEG
P0083	Tunisia	PrMeA	NEG
P0084	Tunisia	TD	NEG
P0085	Algeria	MeA	NEG
P0086	Tunisia	MeA	NEG
P0087	Tunisia	SCOS and PoMeA	NEG
P0088	Tunisia	MeA	NEG
P0089	Tunisia	SCOS	NEG
P0090	Tunisia	SCOS	NEG
P0091	Tunisia	SCOS	NEG
P0092	Algeria	SCOS	NEG
P0093	Algeria	PoMeA	NEG
P0109	Algeria	MeA	NEG
P0110	Tunisia	MeA	NEG
P0111	Tunisia	SCOS	NEG
P0112	Algeria	SCOS	NEG
P0113	Algeria	SCOS	NEG
P0133	Tunisia	SCOS	POS
P0134	Algeria	HS	POS
P0135	Algeria	TD	POS
P0136	Algeria	TD	POS
P0137	Algeria	PrMeA	NEG
P0138	Algeria	TD	NEG
P0139	Algeria	TD	NEG
P0140	Algeria	SCOS	POS
P0141	Algeria	SCOS	NEG
P0142	Algeria	MeA	NEG
P0143	Tunisia	SCOS	NEG
P0144	Tunisia	MeA	NEG
P0145	Algeria	MeA	NEG
P0195	Tunisia	SCOS	NEG
P0196	Algeria	TD	NEG
P0202	Algeria	MeA	NEG
P0203	Tunisia	MeA	NEG
P0242	Algeria	PrMeA	NEG
P0243	Tunisia	MeA	NEG
P0252	Algeria	PoMeA	NEG
P0253	Tunisia	HS	POS

P0254	Tunisia	TD	POS
P0255	Tunisia	HS and TD	POS
P0256	Lybia	HS	POS
P0257	Tunisia	PoMeA	NEG
P0258	Tunisia	HS	POS
P0259	Tunisia	HS	POS
P0260	Algeria	TD	NEG
P0270	Tunisia	TD	POS
P0271	Algeria	HS	POS
P0272	Algeria	HS	POS
P0273	Tunisia	SCOS	POS
P0274	Tunisia	MeA	POS
P0275	Tunisia	SCOS	NEG
P0276	Tunisia	TD	NEG
P0277	Algeria	SCOS	NEG
P0278	Tunisia	MeA	NEG
P0279	Tunisia	MeA	NEG
P0280	Tunisia	TD	NEG
P0281	Tunisia	SCOS	NEG
P0282	Tunisia	SCOS	NEG
P0283	Tunisia	PoMeA	NEG
P0321	Tunisia	MeA	NEG
P0322	Algeria	SCOS	NEG
P0323	Algeria	PoMeA	POS
P0324	Tunisia	SCOS	NEG
P0325	Tunisia	HS and TD	POS
P0326	Algeria	MeA	NEG
P0327	Tunisia	PoMeA	POS
P0328	Algeria	SCOS	NEG
P0349	Tunisia	SCOS	NEG
P0350	Tunisia	MeA	NEG
P0351	Tunisia	SCOS	NEG
P0352	Algeria	MeA	POS
P0353	Algeria	SCOS	NEG
P0354	Lybia	SCOS	NEG
P0355	Tunisia	MeA	NEG
P0356	Lybia	SCOS	NEG
P0357	Tunisia	SCOS	NEG
P0358	Tunisia	HS	POS
P0359	Lybia	PrMeA	NEG
P0360	Algeria	HS	POS
P0361	Algeria	SCOS	NEG
P0362	Tunisia	SCOS	NEG
P0363	Mauritania	SCOS and TD	NEG
P0364	Algeria	PrMeA	POS
P0365	Tunisia	HS	POS

P0366	Tunisia	SCOS	NEG
P0368	Algeria	MeA	NEG
P0369	Tunisia	MeA	NEG
P0370	Tunisia	SCOS	NEG
P0394	Tunisia	SCOS	NEG
P0400	Tunisia	SCOS	NEG
P0408	Tunisia	PrMeA	NEG
P0433	Algeria	HS	POS

Supplementary table 2: List of all nucleotide primers used in the study.

Gene name	Primer sequence (5'-3')	Tm (°C)	Product length (bp)
C14ORF39_EX4-F	TCTGGCTCTAGGATCAGGCC	61.4	500
C14ORF39_EX4-R	AGTTCTATTTCTTCTGATCCTCCT	60.1	
DDX25_EX10-F	CATGGTGTCAAGCACAGC	59.4	539
DDX25_EX10-F	ACGAGCATTATGTCAGGTGGA	57.9	
DMC1_EX6-F	GCTGATCCACTTGCCCATACA	60.41	490
DMC1_EX6-R	TGAAAAGTGTTGATAAGACCCATCT	58.46	
DMC1_EX13-F	AGCAGCTCCAAAACCTCACCT	59.52	502
DMC1_EX13-R	AACACACAGCATCTGGCAGT	60.18	
FANCM_EX22-F	AGGCGCTCATAGTTACCCCT	60.4	488
FANCM_EX22-R	ACCTGTTAGCCATCCTTTTCACA	60.18	
GCNA_EX10-F	AAACCACGGGCTTTACACCT	59.82	482
GCNA_EX10-R	ACCTGTACCAGGGCTCATCT	59.96	
HENMT1_EX4-F	ACTCCACTCTCCTCCTCA	59.4	555
HENMT1_EX4-R	GTTCAAATGCAGGCGGTGG	59.4	
HENMT1_EX6-F	TGCCTGGCCGAATAATTGT	57.3	518
HENMT1_EX6-R	TGCCAAATGCAATGGTTCC	57.3	
HFM1_EX32-F	TAGGAACGACTGCCAGCAAA	57.3	560
HFM1_EX32-R	TTCTTAGTCAAGCCACAAGT	55.9	
MCM8_EX5-F	TGGCACAAAGCTTATACTTATGTCA	58.1	568
MCM8_EX5-R	ATTTCTGCACAATCTTTAGCAAGA	55.9	

MCMDC2_EX14-F	AAAACCTTCGGGAGGGGAGTG	59.4	521
MCMDC2_EX14-R	TGCCCAAATTAACAGGCCT	55.9	
MEIOB_EX12-F	CAGAGGACTGGTGTACGCAG	61.4	528
MEIOB_EX12-R	TCCCAGGCTGTTTATTAGTGAA	56.5	
MSH5_EX8-F	TCAGGCTGTGCTTTCAGACA	59.53	419
MSH5_EX8-R	AGGCCAGTACCTCCAGAAT	59.96	
PDHA2_F	TCTGTCCGATCCATTCTCGC	59.61	380
PDHA2_R	CCTATCGACCTTTAGCCCAGG	60.18	
REC8_EX12-F	GGGGCTTTTCTGGGAGTACC	61.4	501
REC8_EX12-R	CAAGGCTGCCATTACTCCT	59.4	
TDRD9_EX5-F	TTAGATGAGCCAACCCCTGC	59.4	533
TDRD9_EX5-R	CAAACACAACATTCACCA	57.9	
TDRD9_EX30-F	TGAAGAGAATGAGTTACATCCAGA	59.28	438
TDRD9_EX30-R	CTGCACTCCAGCCTGAATGA	60.03	
TDRKH_EX7-F	TGTACCTCTCACTACCCTGGT	59.8	519
TDRKH_EX7-R	TTCCATTCTCCAAGGTGCC	59.4	

Supplementary table 3: Bibliographic information on the 151 NOA candidate genes.

See the online table:

<https://docs.google.com/spreadsheets/d/14uA4h43deSiwqdrKk81JYPOjnh-E0UI-jK9ntkyburg/edit?usp=sharing>

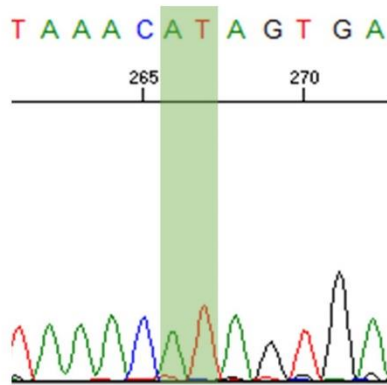
Supplementary Figure 1: Sanger sequencing confirmation of all identified gene variants.

Upper panels: Electrophoregrams from Sanger sequencing showing the identified variants. The substituted/deleted nucleotides are highlighted in green.

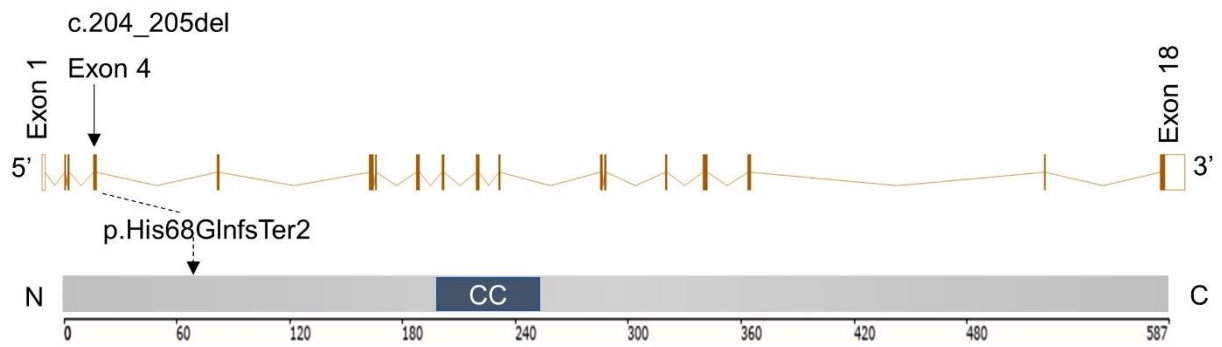
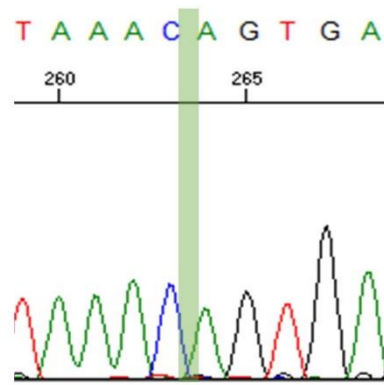
Lower Panel: Location of these variants within each gene and protein represented with domains (according to UniprotKB).

C14orf39 ENST00000321731.7

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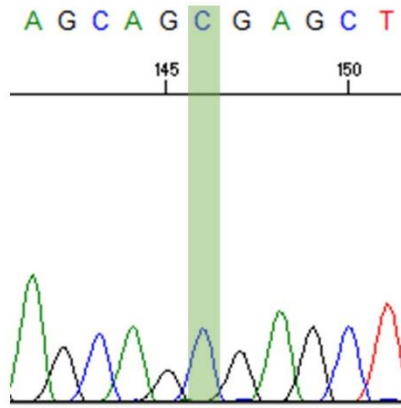


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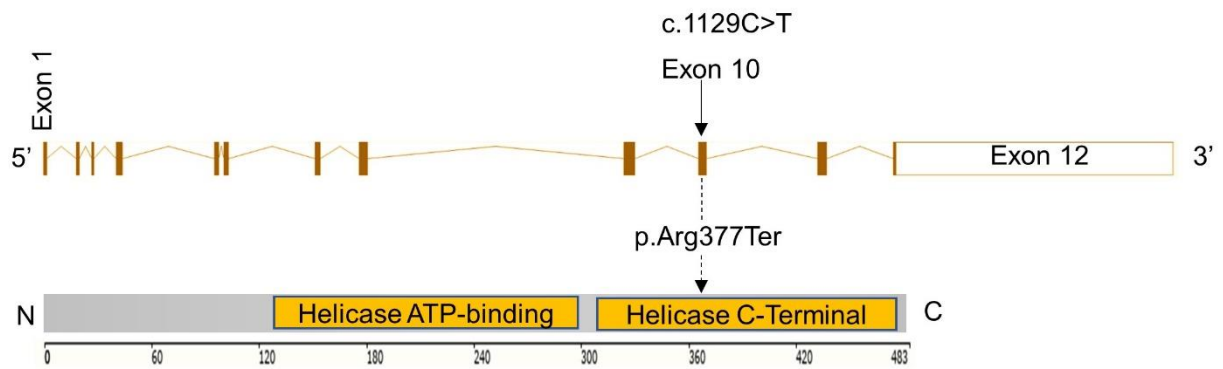
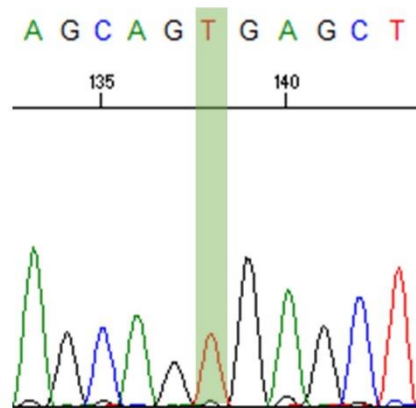


DDX25 ENST00000263576.10

Control



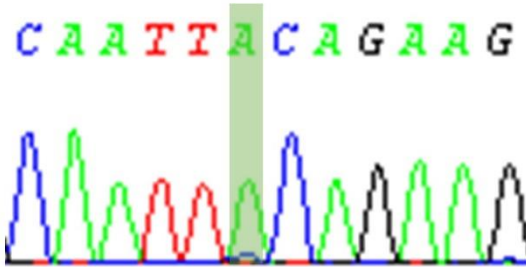
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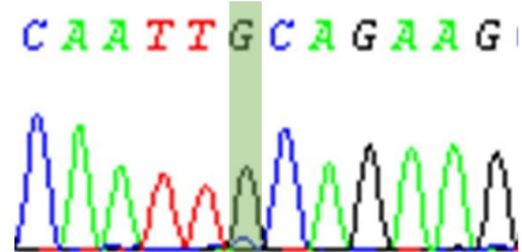
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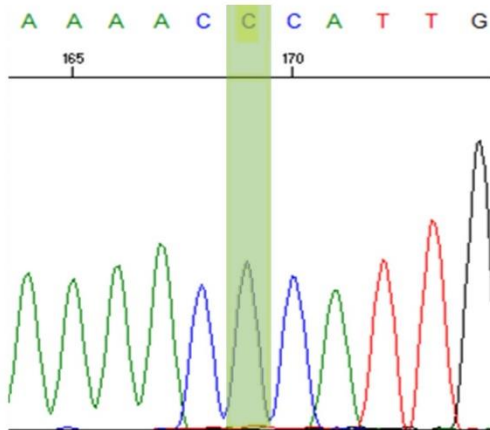


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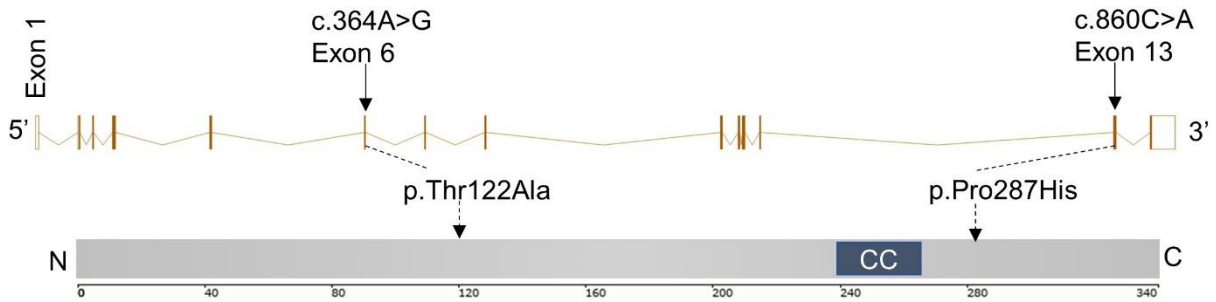
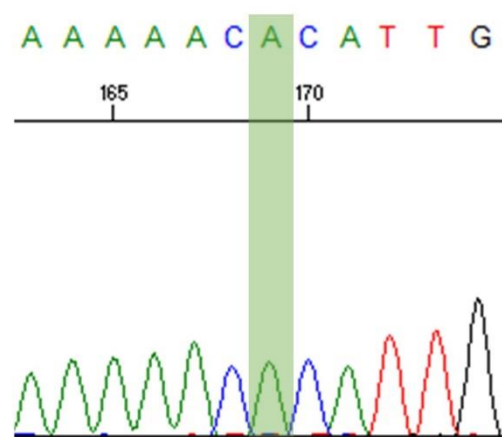


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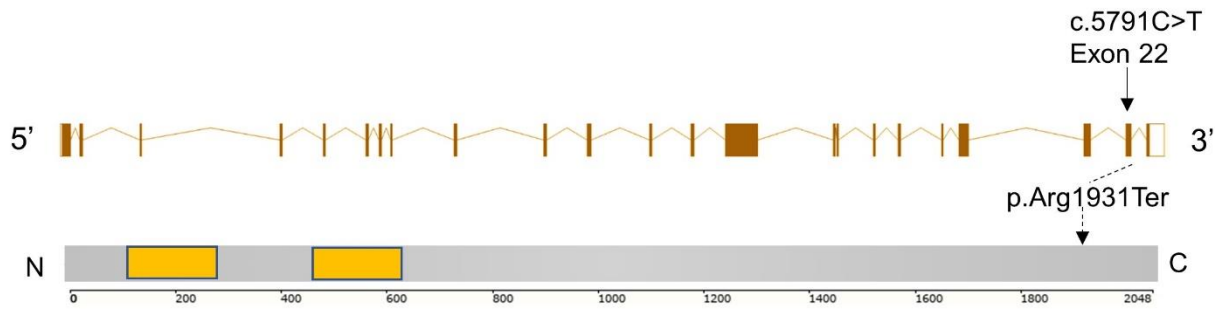
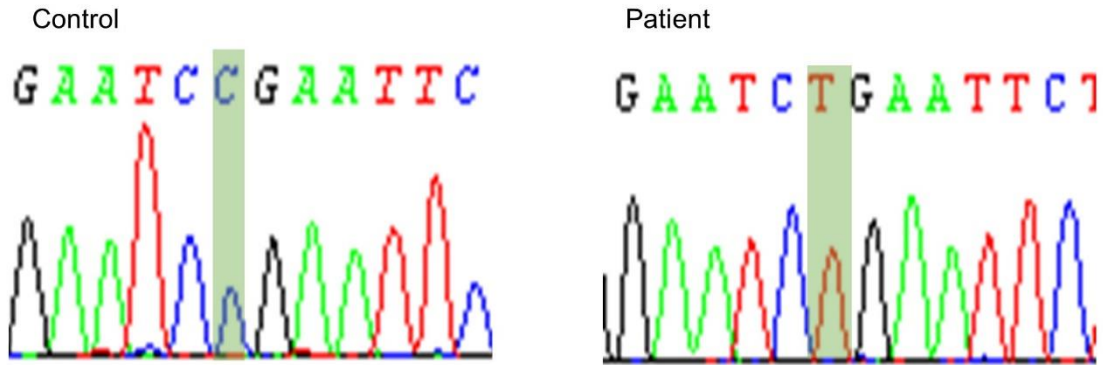
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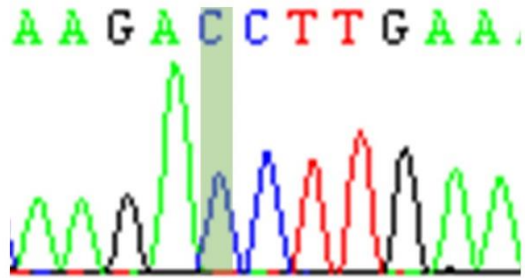


FANCM ENST00000267430.9

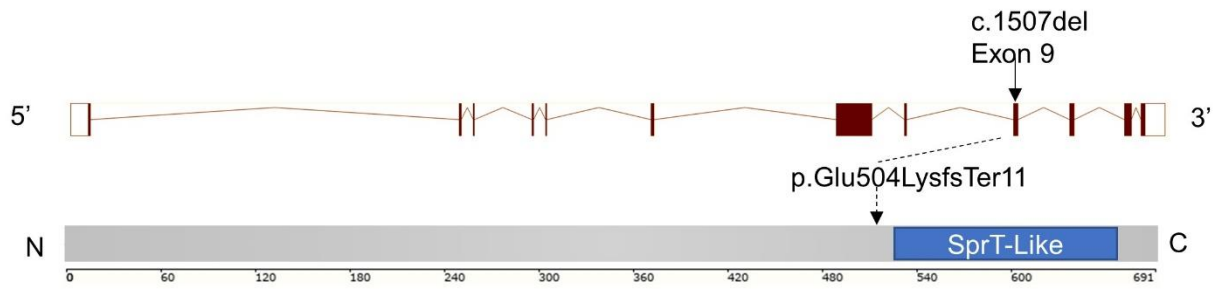
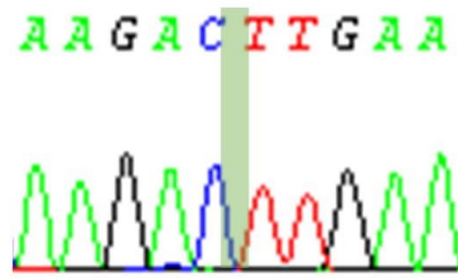


GCNA ENST00000373695.1

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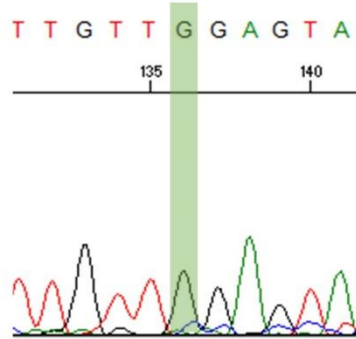
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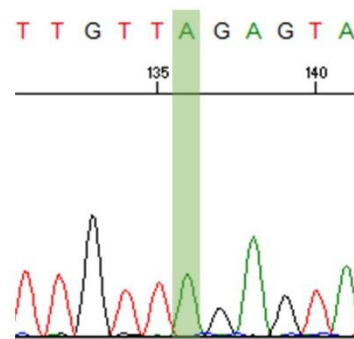
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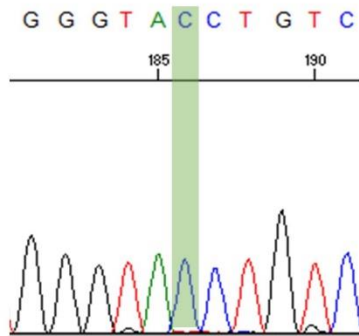


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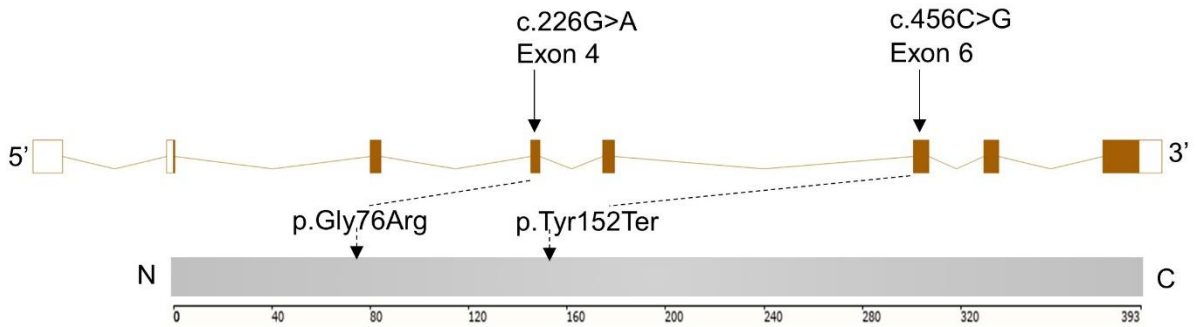
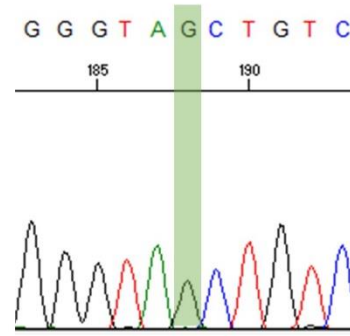


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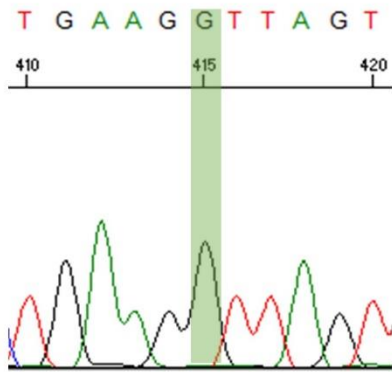


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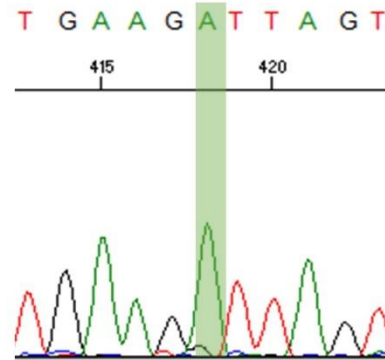


HFM1 ENST00000370425.7

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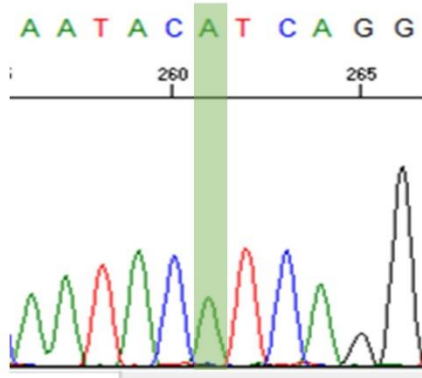


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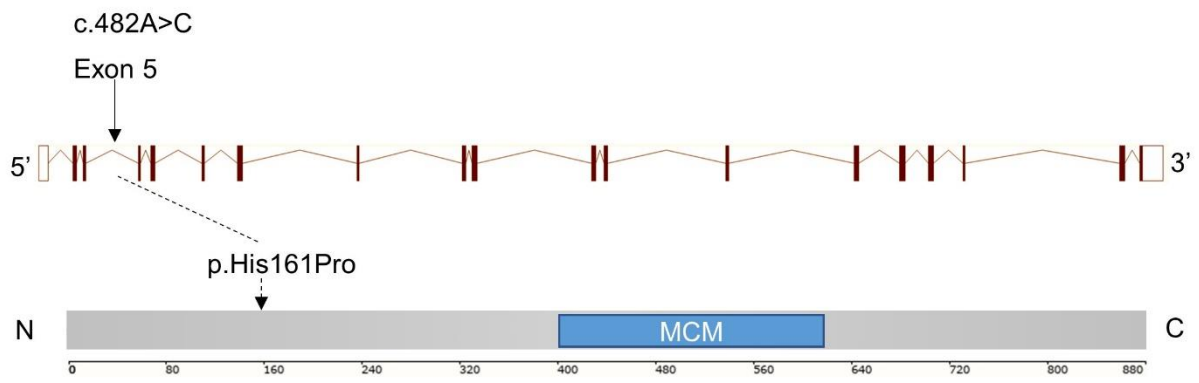
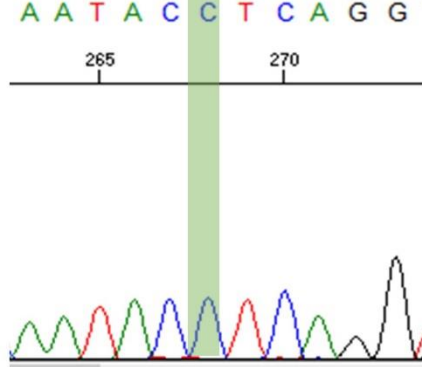


MCM8 ENST00000378886.6

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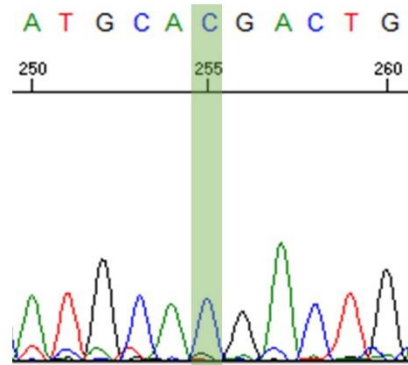


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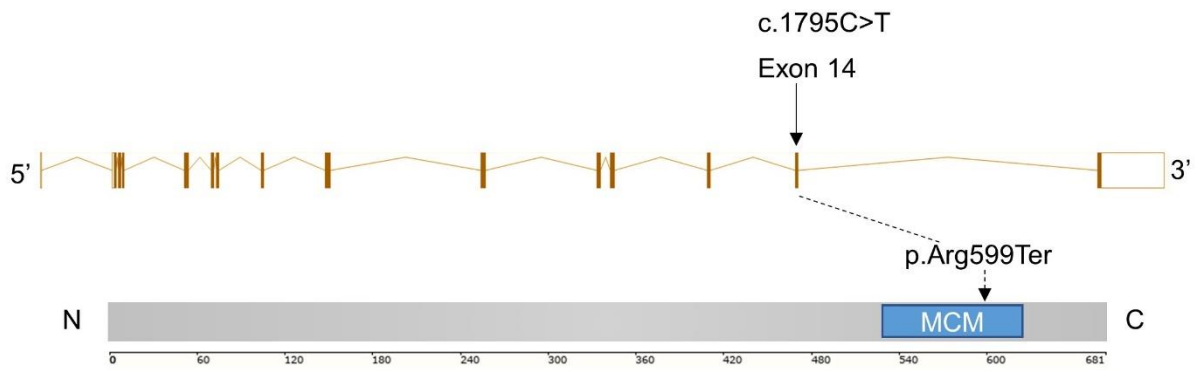
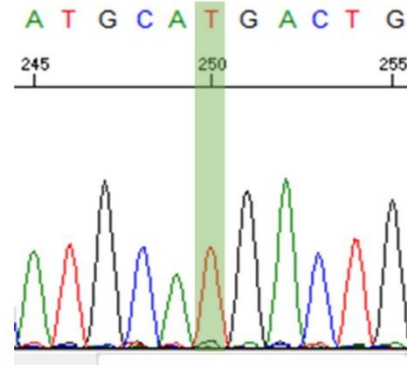


MCMD2 ENST00000422365.6

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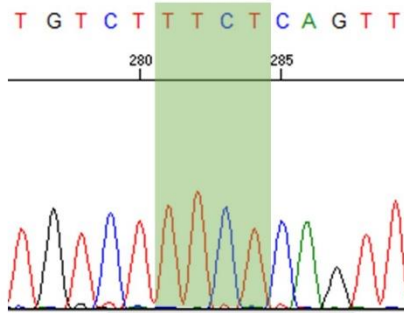


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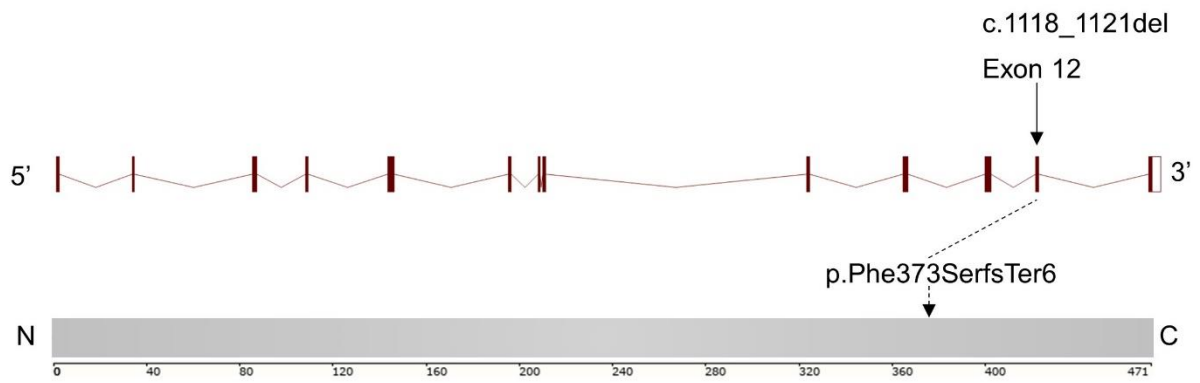
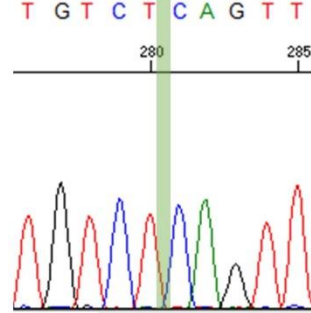


MEIOB ENST00000325962.7

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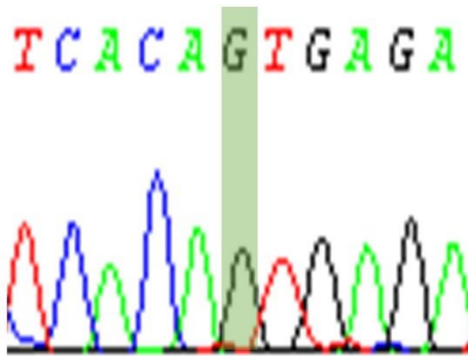


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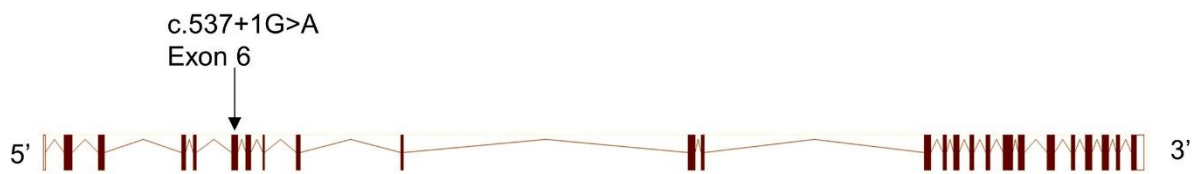
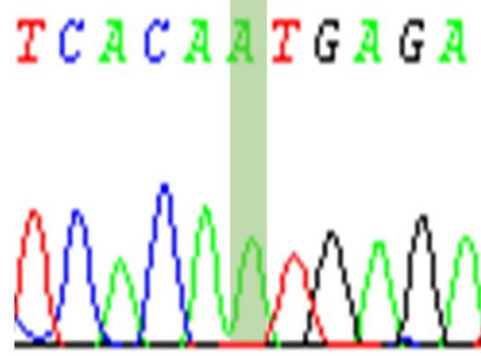


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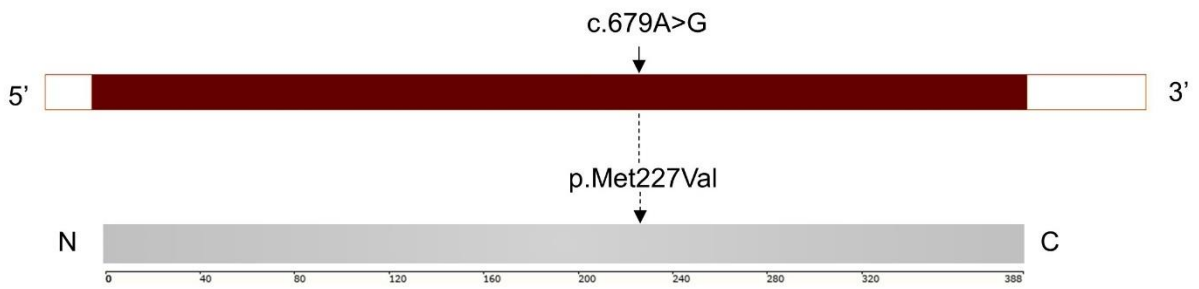
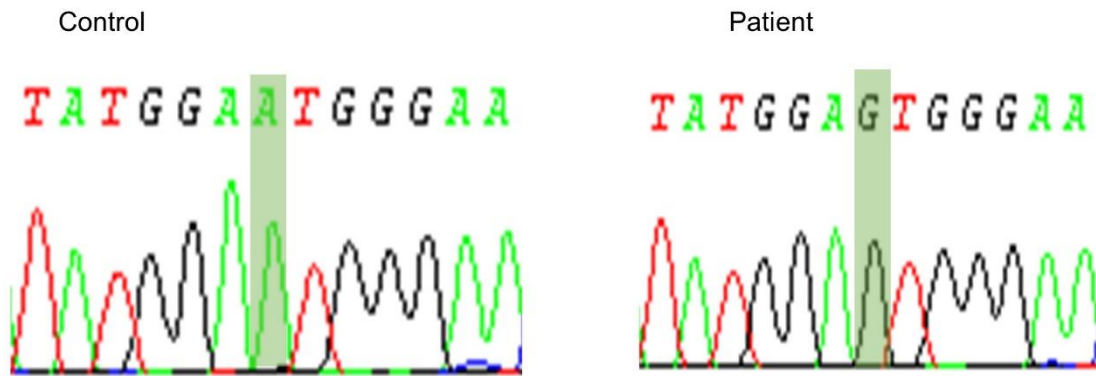
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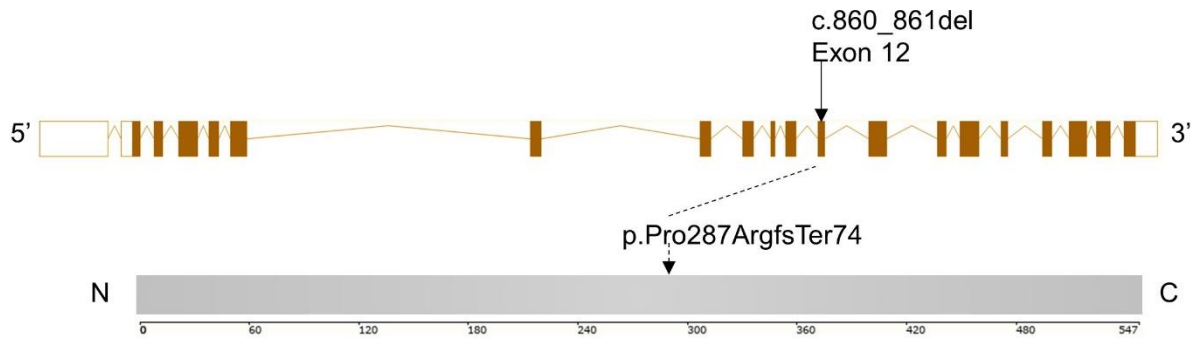
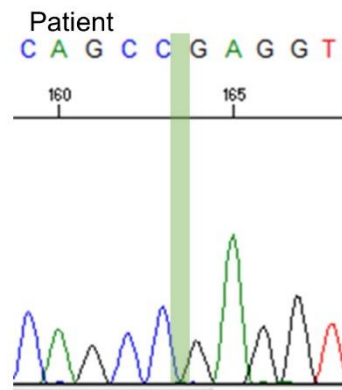
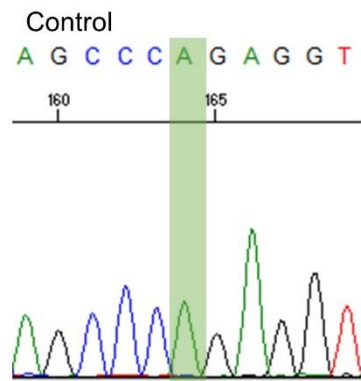
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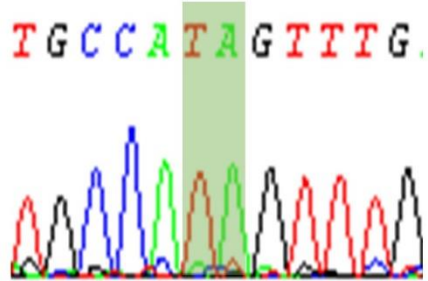
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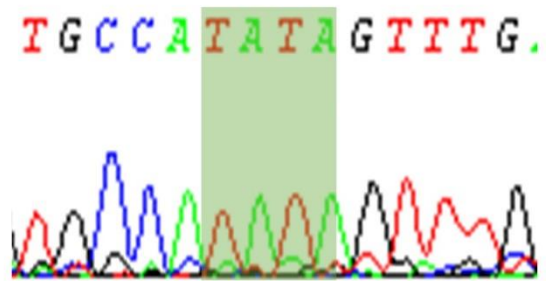
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Exon 30

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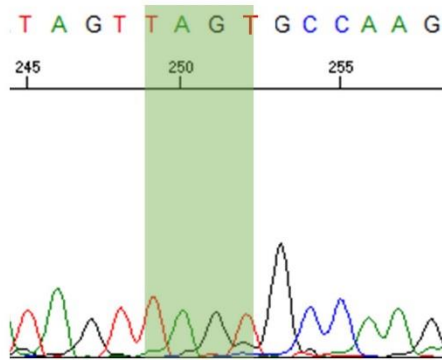


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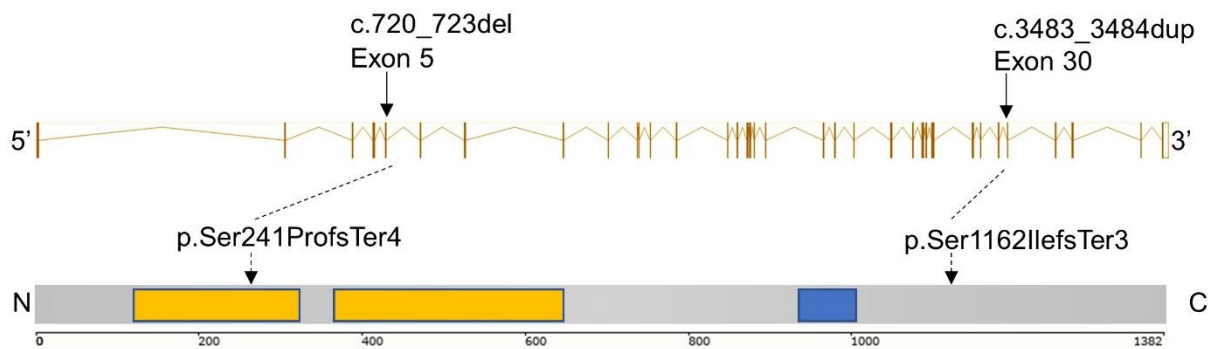
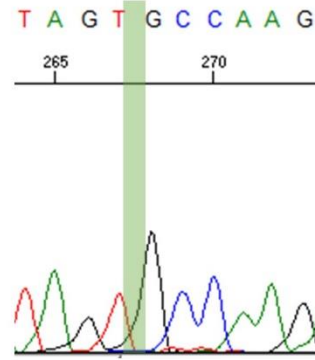


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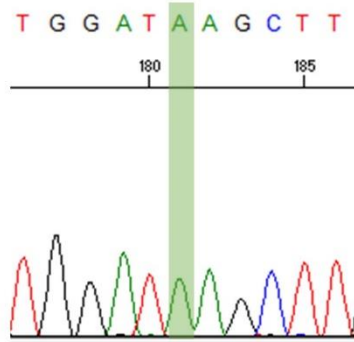


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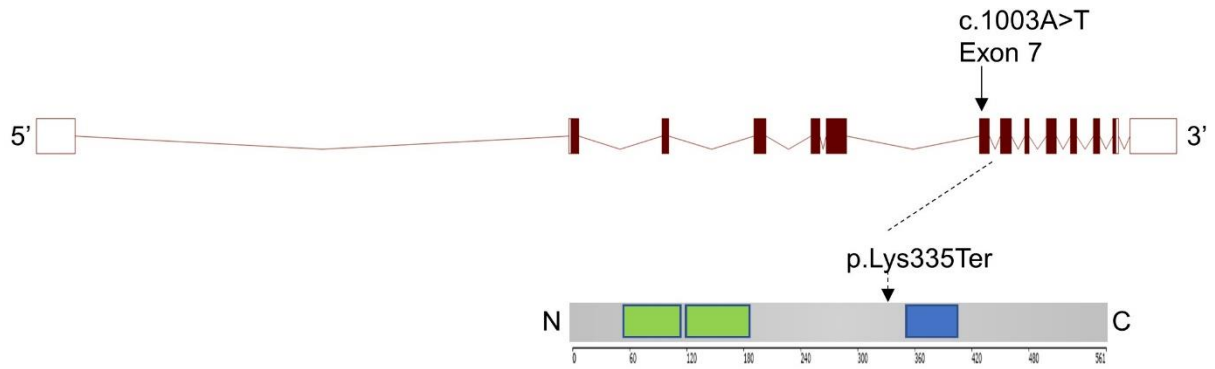
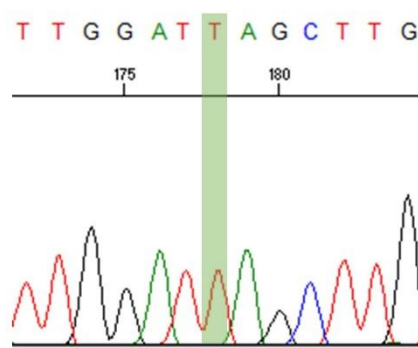


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Control



Patient



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