Reproductive Biology and Endocrinology

CORRECTION

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Correction: Identification of deleterious variants in patients with male infertility due to idiopathic non-obstructive azoospermia

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Following the publication of the original article [1], it was noted that due to a typesetting error the figure images for Figures 1-5 in the PDF version were not updated and an error was found in Table 1.

The correct Figs. 1, 2, 3, 4, 5 and Table 1 are shown below.

The original article [1] has been corrected.

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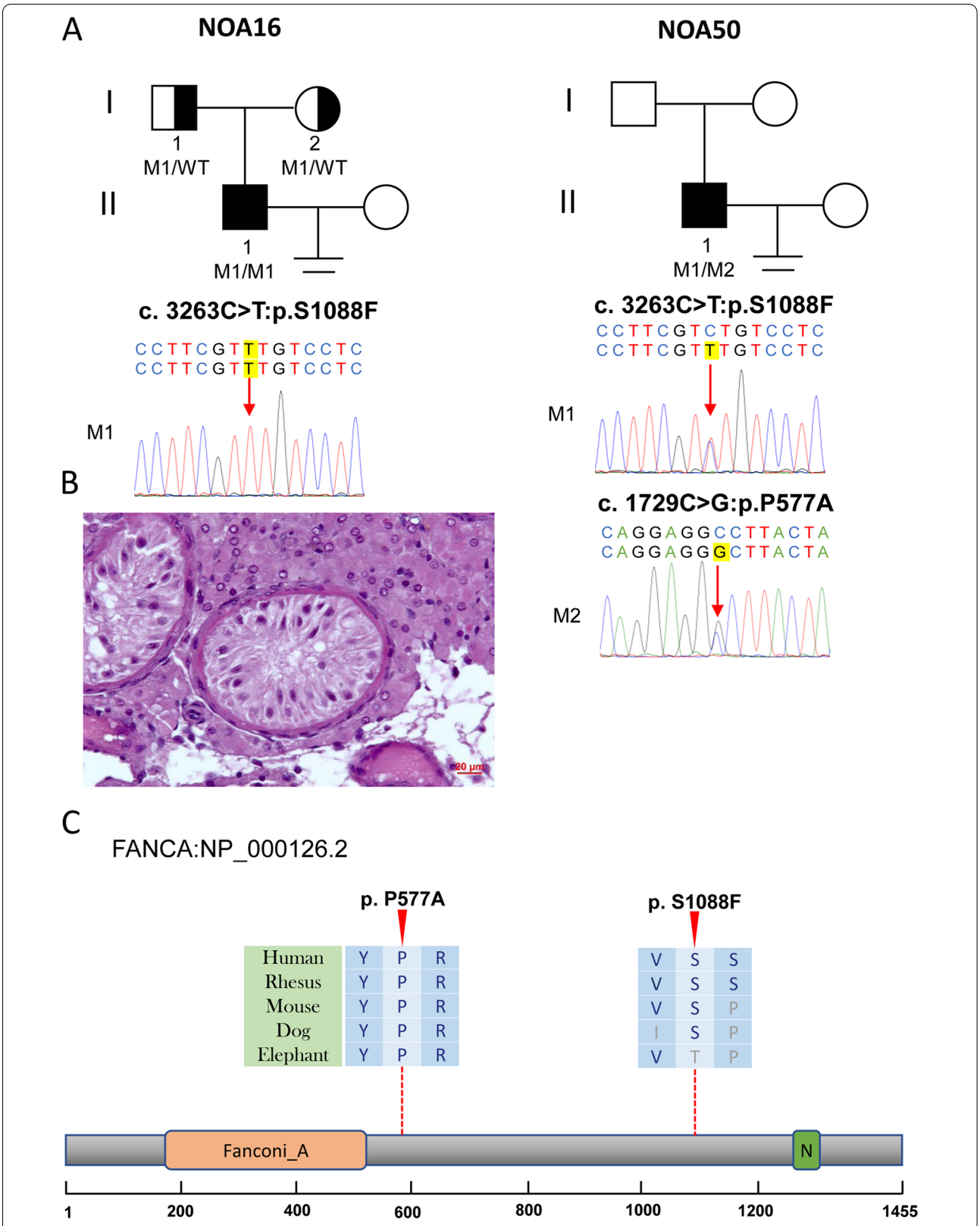

Fig. 1 Variants of *FANCA* in NOA16 and NOA50. **A** The families affected by the variants in *FANCA*. The red dotted lines indicate mutated positions in the Sanger sequencing results. **B** Testicular histopathology of NOA16. **C** The mutated positions of *FANCA* are conserved among species (red arrows). And the dotted lines indicate the positions of the *FANCA* variant in the *FANCA* protein. M, mutation; WT, wild type

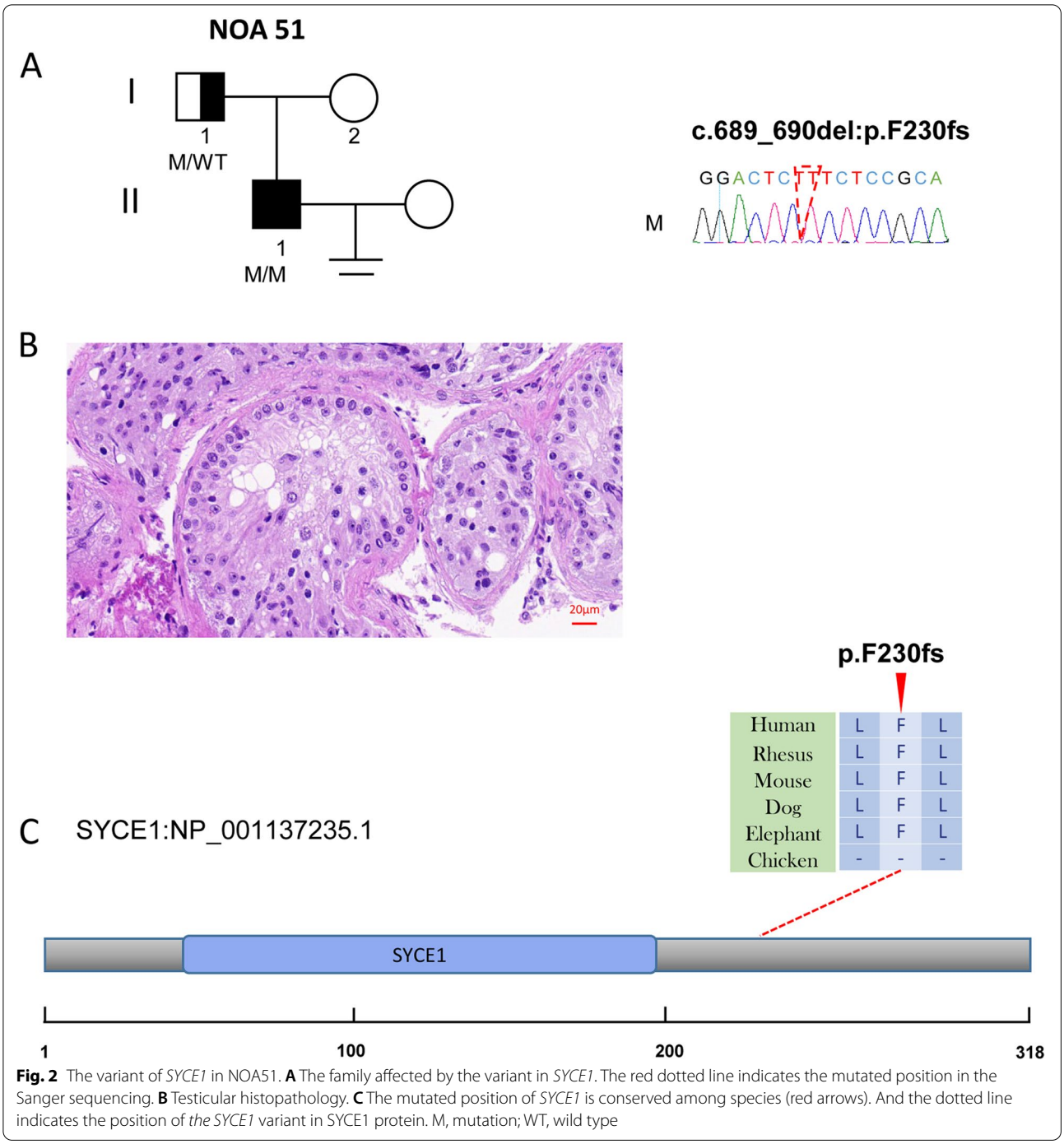

Fig. 2 The variant of *SYCE1* in NOA51. **A** The family affected by the variant in *SYCE1*. The red dotted line indicates the mutated position in the Sanger sequencing. **B** Testicular histopathology. **C** The mutated position of *SYCE1* is conserved among species (red arrows). And the dotted line indicates the position of *the SYCE1* variant in SYCE1 protein. M, mutation; WT, wild type

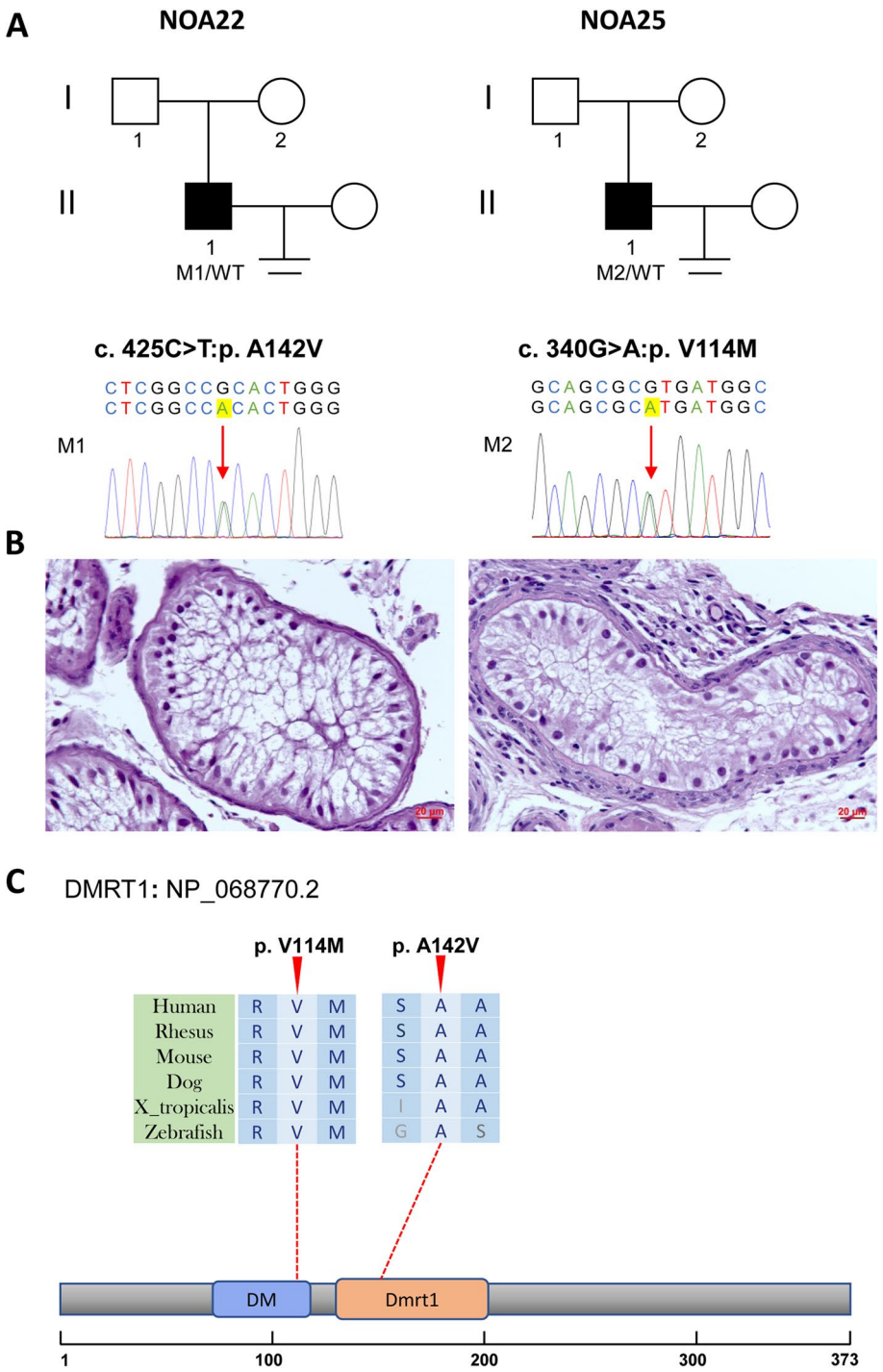

Fig. 3 Variants of *DMRT1* in NOA22 and NOA25. **A** The families affected by the variants in *DMRT1*. The red arrows indicate mutated positions in the Sanger sequencing results. **B** Testicular histopathology. **C** The mutated positions of *DMRT1* are conserved among species (red arrows). And the dotted line indicates the position of *DMRT1* variants in *DMRT1* protein. M, mutation; WT, wild type

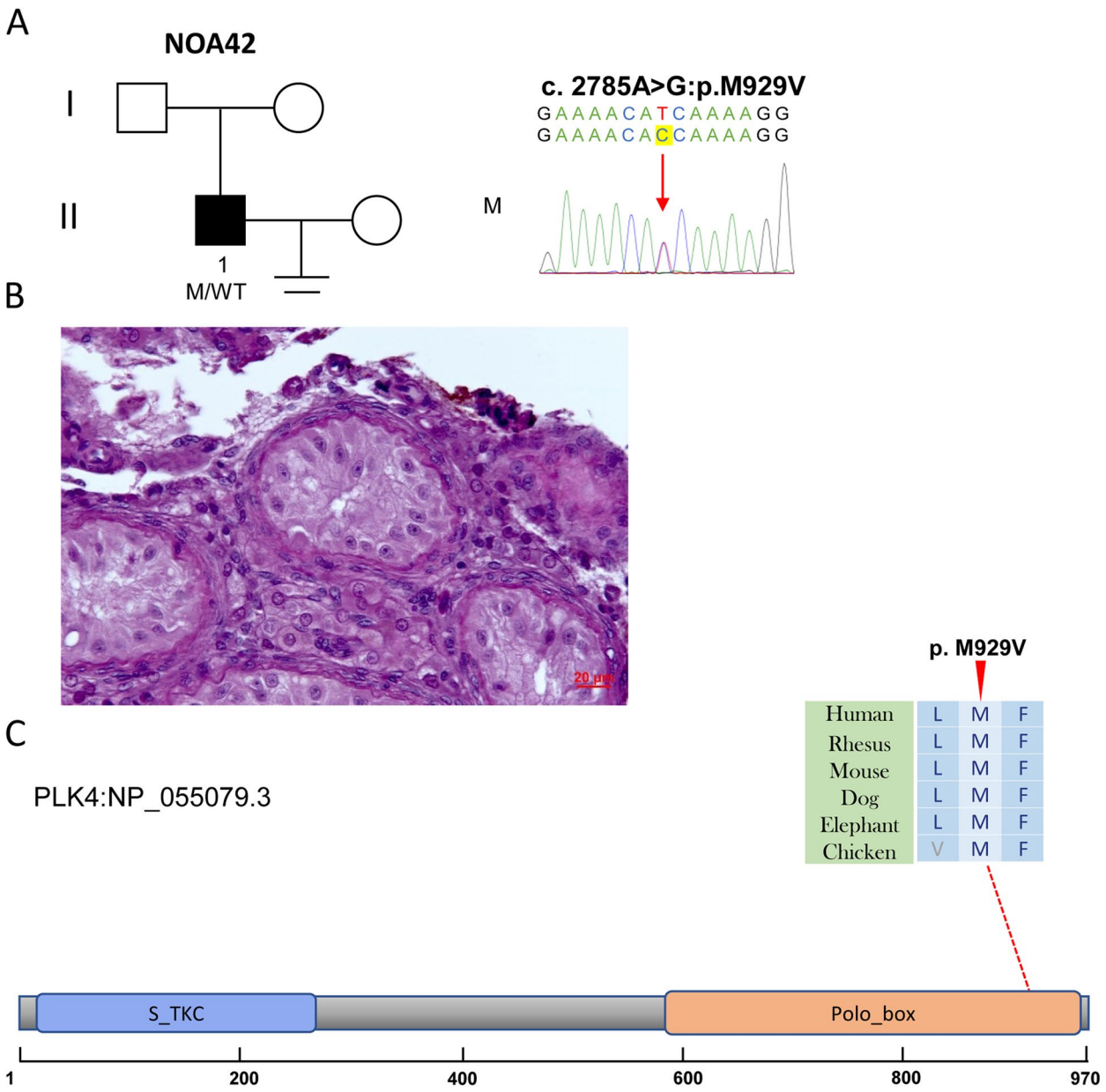

Fig. 4 The variant of *PLK4* in NOA42. **A** The family affected by the variant in *PLK4*. The red arrow indicates the mutated position in the Sanger sequencing results. **B** Testicular histopathology. **C** The mutated position of *PLK4* is conserved among species (red arrows). And the dotted line indicates the position of *PLK4* variant in PLK4 protein. S_TKC, Serine/Threonine protein kinases, catalytic domain; M, mutation; WT, wild type

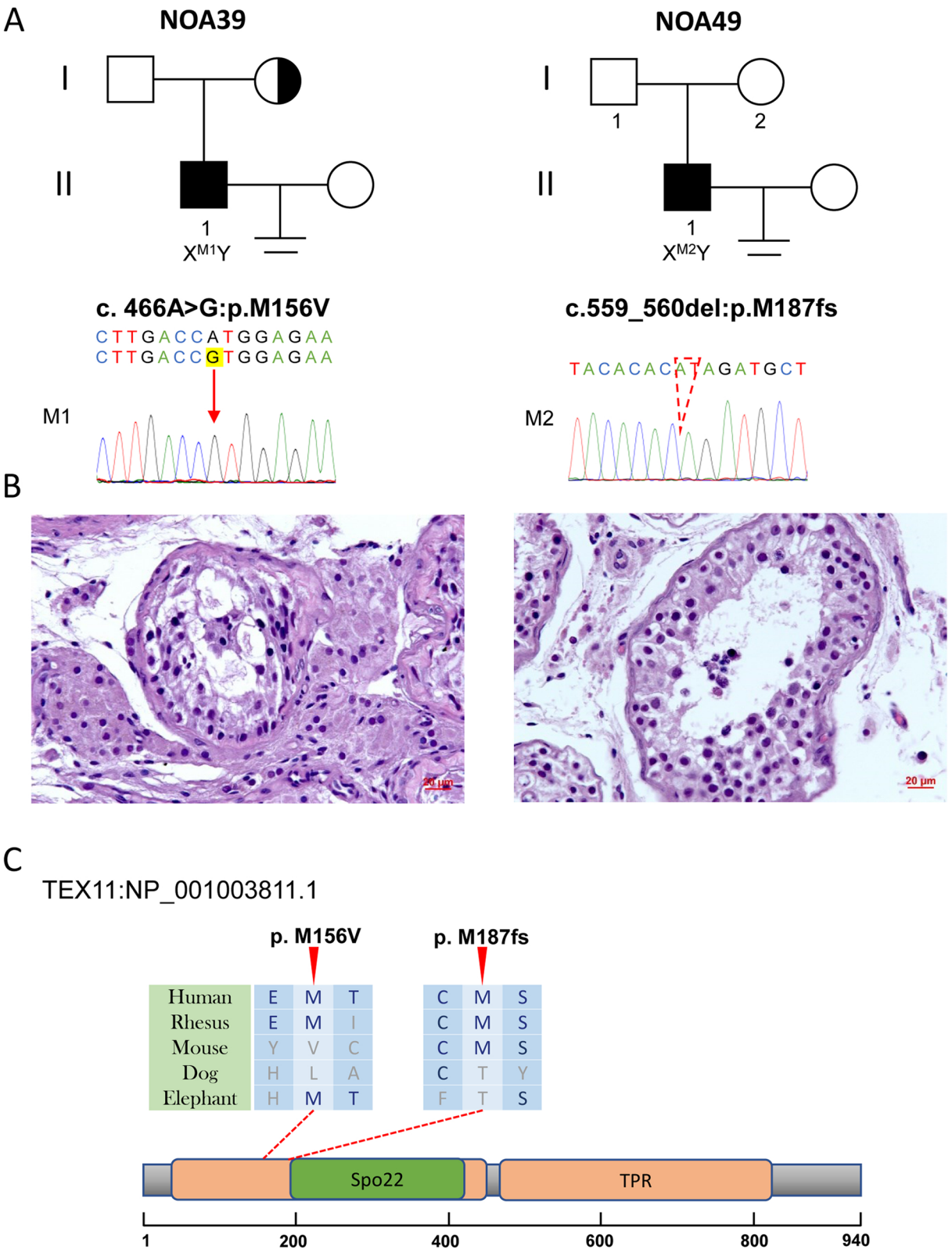

Fig. 5 Variants of *TEX11* in NOA39 and NOA49. **A** The families affected by the variants in *TEX11*. The red dotted line indicates mutated positions in the Sanger sequencing results. **B** Testicular histopathology. **C** The mutated positions of *TEX11* are conserved among species (red arrows). And the dotted lines indicate the positions of *TEX11* variants in TEX11 protein. M, mutation; WT, wild type

Table 1 Deleterious variants detected in patients with non-obstructive azoospermia and related clinical phenotypes.

Individual	NOA16	NOA50	NOA51	NOA22	NOA25	NOA42	NOA39	NOA49
Gene	*FANCA*	*FANCA*	*SYCE1*	*DMRT1*	*DMRT1*	*PLK4*	*TEX11*	*TEX11*
Inheritance pattern	AR	AR	AR	AD	AD	AD	X-linked	X-linked
RefSeq accession number	NM_000135	NM_000135	NM_001143763	NM_021951	NM_021951	NM_001190799	NM_031276	NM_031276
Age	27	27	31	27	31	29	32	25
Secondary sexual characteristics	Normal	Normal	Normal	Normal	Normal	Normal	Normal	Normal
testicular volume (Left/Right, ml)	8/8	8/8	15/15	10/10	10/10	12/12	12/12	10/10
Somatic karyotype	46,XY	46,XY	46,XY	46,XY	46,XY	46,XY	46,XY	46,XY
Y Chromosome microdeletions	No	No	No	No	No	No	No	No
Follicle-stimulating hormone (IU/L)	23.87	24.74	3.85	16.32	26.54	29.24	8.44	4.02
Luteinizing hormone (IU/L)	6.10	9.38	0.41	6.44	11.35	7.05	6.33	5.33
Testosterone (nmol/L)	14.03	9.64	31.14	17.95	7.07	10.75	10.75	13.34
Estradiol (pmol/L)	NA	90	372	241	23	73	97	132
Prolactin (ng/ml)	NA	8.26	14.6	11.88	10.37	8.11	8.92	10.24
Testis histology	SCOS	ND	MA	SCOS	SCOS	SCOS	Hypospermatogenesis	MA
Hom/Het	Hom	Het/ Het	Hom	Het	Het	Het	Hemi	Hemi
cDNA mutation	c.3263C>T	c.3263C>T/ c.1729C>G	c.689_690del	c.425C>T	c.340G>A	c.2785A>G	c.466A>G	c.559_560del
Mutation type	Missence	Missence/ Missense	Frameshift	Missense	Missense	Missense	Missense	Frameshift
Protein alteration	p.S1088F	p.S1088F/ p.P577A	p.F230fs	p.A142V	p.V114M	p.M929V	p.M156V	p.M187fs
1KGP	0.0218	0.0218/ 0	0	0	0	0	0	0
EXAC_EAS	0.0235	0.0235/ 0	0	0	0	0	0.0039	0
gnomAD_EAS	0.0265	0.0265/ 0	0.0001	0	0	0	0.0034	0
SIFT	D	D/ D	NA	T	D	D	T	NA
PolyPhen-2	P	P/ D	NA	D	D	P	B	NA
MutationTaster	N	N/ D	NA	D	D	D	N	NA
CADD	21.8	21.8/ 23.9	NA	22.2	33	23.9	22.2	NA
HGMD	NA	NA/ NA	NA	NA	NA	NA	D	NA
Validation in patient	Yes	Yes/ Yes	Yes	Yes	Yes	Yes	Yes	Yes
Mother/Father genotype	Het/Het	ND/ ND	ND/Het	ND	ND	ND	Het/WT	ND

AR autosomal recessive, *AD* autosomal dominant, *1KGP* 1000 Genomes Project, *ExAc_EAS* the data of East Asian in Exome Aggregation Consortium, *gnomAD_EAS* the data of East Asian in the Genome Aggregation Database, *D* Damaging, *T* Tolerant, *P* Possibly Damaging, *B* Benign, *N* Polymorphism, *ND* Not Detect, *SCOS* Sertoli cell only syndrome, *MA* maturation arrest