

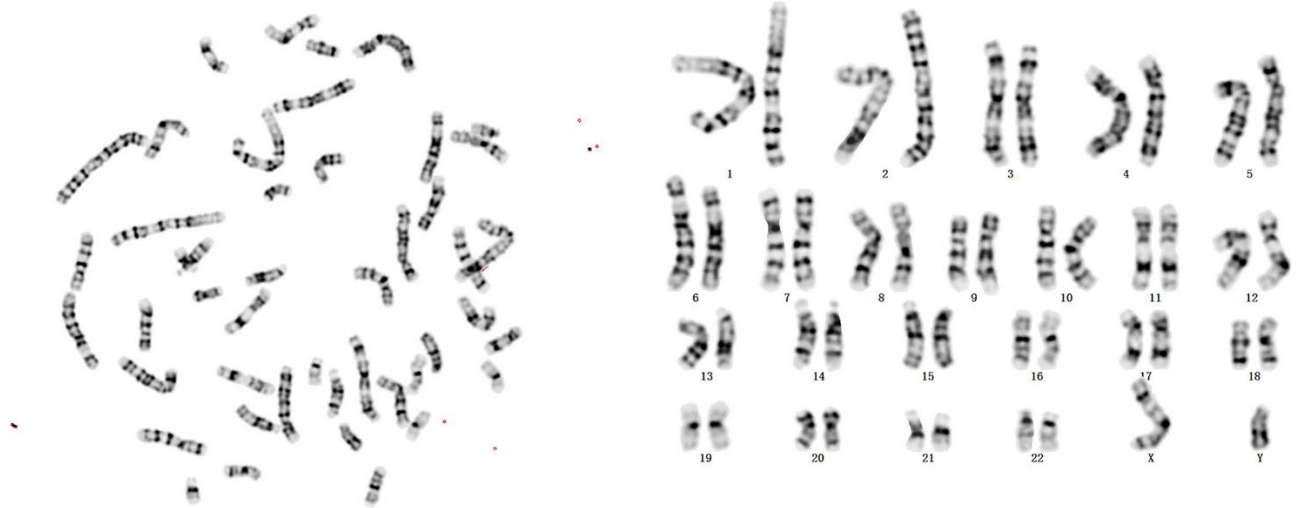
1 **A loss-of-function variant in *SSFA2* causes male infertility with**
2 **globozoospermia and failed oocyte activation**

3 *Huang et al.*

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5 **Supplementary information**

6 **Supplementary Figures**



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8 **Supplemental Figure S1. Karyotype analysis of the patient: normal male karyotype 46, XY.**

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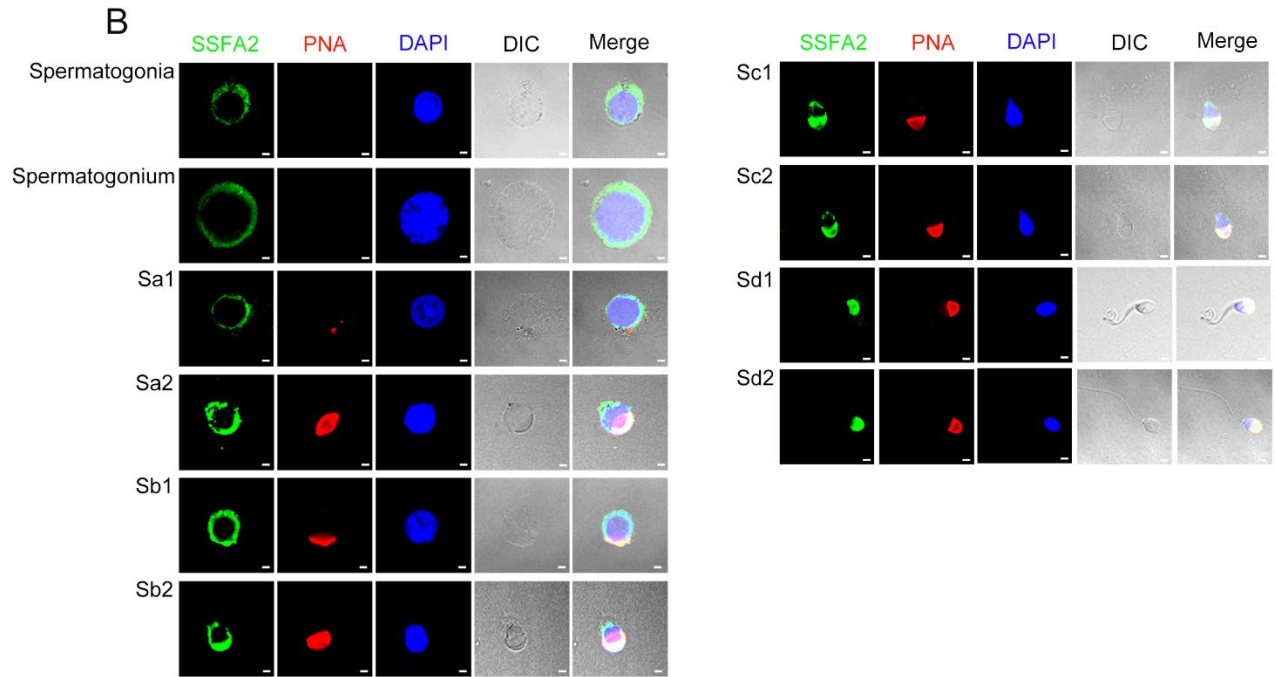
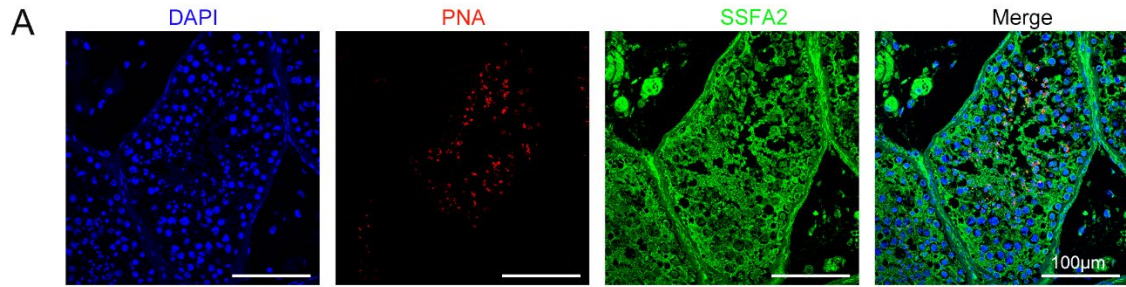
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19 **Supplemental Figure S2. The expression pattern of SSFA2 in humans.**

20 (A) SSFA2 showed higher expression in early and late spermatids, whereas it was also expressed
 21 in spermatogonia and spermatocytes (green, SSFA2; red, PNA; blue, DAPI; scale bars, 100 µm).

22 (B) The expression pattern of SSFA2 in spermatogenic cells at different stages. Sa1-Sb1, round
 23 spermatid. Sb2-Sd2, elongating spermatids (green, SSFA2; red, PNA; blue, DAPI; scale bars, 1
 24 µm).

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26 **Supplementary tables**

27 **Supplemental Table SI. Basal blood hormone levels**

Hormone	Baseline	Reference Range	28
E2 (pmol/L)	41.0	0-156	29
FSH (mIU/mL)	3.2	1.5-12.4	
LH (mIU/mL)	2.2	1.7-8.8	30
PRL (mIU/L)	283.4	86-324	
Testosterone (nmol/L)	14.2	8.6-29.0	31

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46 **Supplemental Table SII. Analysis of *SSFA2* Variant in the patient**

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Phenotype	Globozoospermia
cDNA mutation	c.3671G>A
Genotype	Homozygous
Protein alteration	p.R1224Q
Mutation type	Missense
dbSNP	0
Allele frequency in ExAC	0
1000G project	0
gnomAD	0.00000427
Function prediction	
SIFT	Deleterious
PolyPhen-2	Probably damaging
CADD_phred	24.1 ^a
Mutation Taster	Disease causing

48 *The GenBank accession number for *SSFA2*: NM_001130445.3

49 ^a The points>10 means deleterious

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