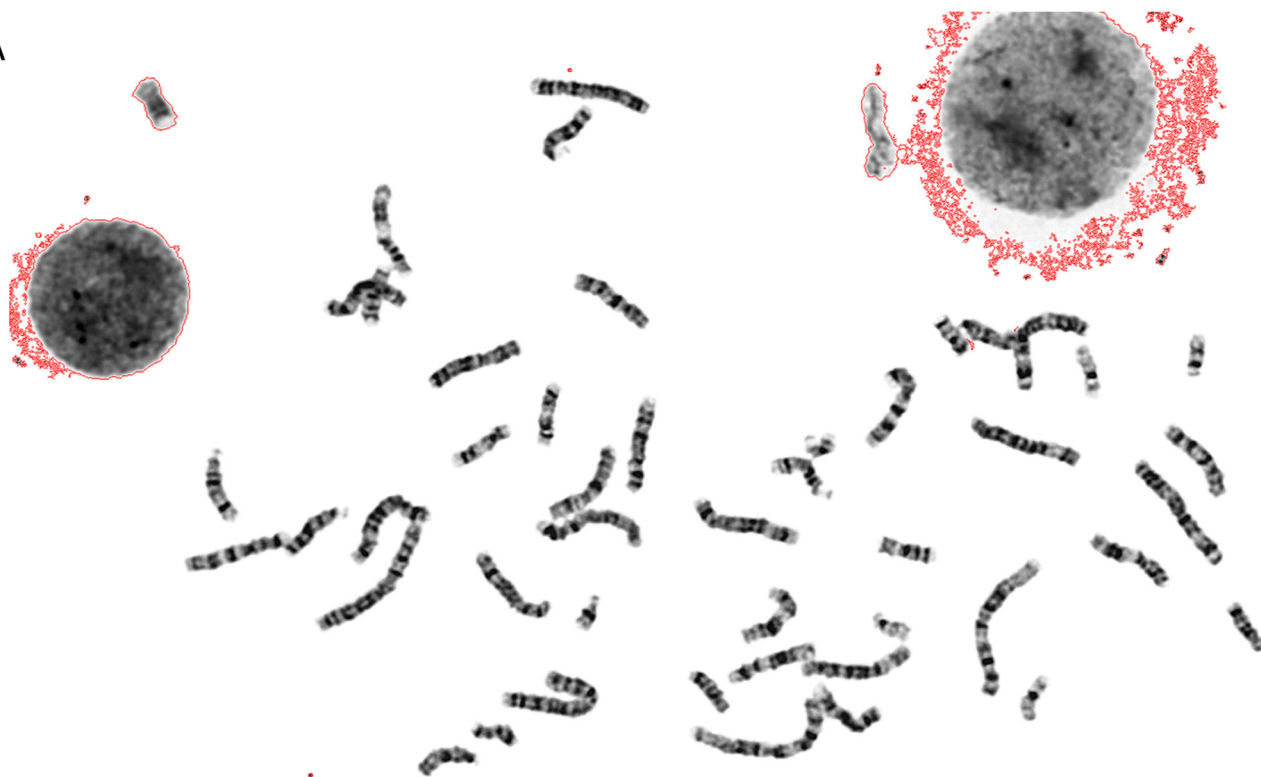


Figure S1. Evidence that excludes Turner syndrome. A total of 33 metaphase cells were analyzed for the patients, all were normal. (A) Chromosome at Metaphase. (B) Karyotype image, the arrow indicates two normal X chromosomes.

A



B

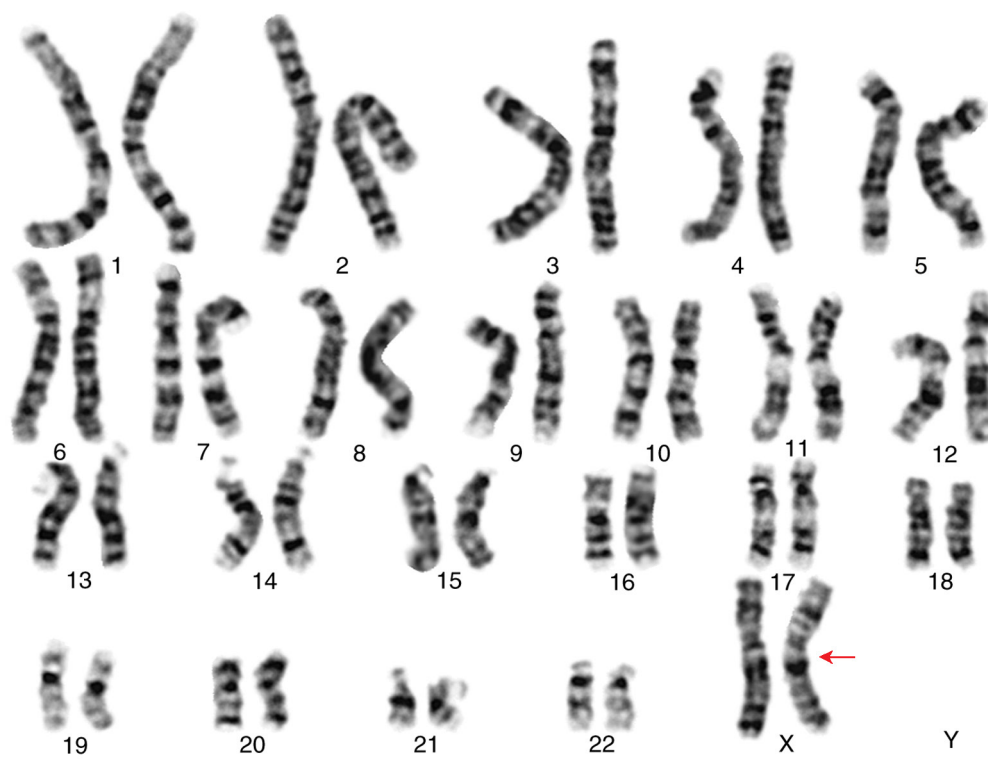


Table SI. POC1 centriolar protein A (NM_015426.5) pathogenic variants reported in humans.

First author/s, year	Population	Families	Type of variant	Pathogenic variant	(Refs.)
Shalev <i>et al</i> , 2012; Sarig <i>et al</i> , 2012	Arab Muslim	2	Homozygous missense	c.512T>C: p.Leu171Pro	(1,3)
Shaheen <i>et al</i> , 2012	Saudi Arabia	3	Homozygous nonsense	c.241C>T:p.Arg81*	(2)
Koparir <i>et al</i> , 2015	Turkish	1	Homozygous missense	c.358A>G:p.Thr120Ala	(5)
Barraza-García <i>et al</i> , 2016	Spanish	1	Homozygous truncating	c.254delT:p.Leu85Alafs*22	(7)
	Moroccan	1	Homozygous truncating	c.515G>A:p.Trp172*	
Ko <i>et al</i> , 2016	Korean	1	Compound heterozygous	c.239C>T:p.Ser80Phe; c.241>T: p.Arg81*	(6)
Mostofizadeh <i>et al</i> , 2018	Iranian	1	Homozygous missense	c.G491A:pSer164Asn	(9)
Saida <i>et al</i> , 2019	Chilean	1	Homozygous missense	c. 649C>T:p.Arg217Trp	(8)
Al-Kindi <i>et al</i> , 2020	Omani	2	Homozygous missense	c.64G>T: p.Val22Phe	(4)
Chen <i>et al</i> , 2015	Italian	1	Homozygous truncating	c.1048delC: p.Gln250Argfs*4	(10)
Giorgio <i>et al</i> , 2017	Italian	1	Homozygous frameshift	c.1047_1048dupC: p.Gly337Argfs*25	(11)
Present study	Chinese	1	Compound heterozygous	c.850_851insG:p.Glu284Glyfs*9; c.593_605delGTGGGACGTG CAT: p.Ser198Metfs*10	-