Supplemental material

Table S1. Summary of studies on the proportion of autosomal dominant cases among males with RP

Study	Geography	N for male autosomal dominant cases	Total classifiable male RP cases	% of male RP cases that are autosomal dominant
Ayuso et al. 1995 (19)	Spain	132	523	25.2%
Berson et al. 1980 (20)	United States	24	259	9.3%
Bunker et al. 1984 (11)	United States (Maine)	36	92	39.1%
Dickinson and Mulhall 1989 (21)	Australia	76	296	25.7%
Haim 2002 (13)	Denmark	35	465	7.5%
Nájera et al. 1995 (17)	Spain	66	149	44.3%
Testa et al. 2014 (22)	Italy	48	319	15.0%

RP, retinitis pigmentosa.

Table S2. Summary of studies on the proportion of simplex cases among males with RP

Study	Geography	N for male simplex cases	Total classifiable male RP cases	% of male RP cases that are simplex
Ayuso et al. 1995 (19)	Spain	116	523	22.2%
Berson et al. 1980 (20)	United States	183	259	70.7%
Bunker et al. 1984 (12)	United States (Maine)	20	92	21.7%
Dickinson and Mulhall 1989 (21)	Australia	122	296	41.2%
Haim 2002 (13)	Denmark	204	465	43.9%
Nájera et al. 1995 (17)	Spain	33	149	22.1%
Testa et al. 2014 (22)	Italy	142	319	44.5%

RP, retinitis pigmentosa.

Table S3. Summary of studies on the proportion of *RPGR* mutations among individuals/families with XLRP, all studies (includes studies with N<20)

Study	Location	Families/ Individuals	Screened for *RPGR*	Number with any XLRP mutation identified	Number with *RPGR* mutation	% with *RPGR* mutation among those with any known mutation for XLRP	% among all genetically evaluated
Bader et al. 2003 (33)	Germany (93%)[a]	Individuals	58	37	34	91.9%	58.6%
Bocquet et al. 2013 (34)	France	Families	26	22	20	90.9%	76.9%
Breuer et al. 2002 (8)	North America	Individuals	185	70	60	85.7%	32.4%
Dan et al. 2020 (35)	China	Individuals	N/A	5	3	60.0%	N/A
Koyanagi et al. 2019 (36)	Japan	Individuals	N/A	26	23	88.5%	N/A
Kurata et al. 2019 (37)	Japan	Families	N/A	12	7	58.3%	N/A
Liu et al. 2021 (31)	China (Beijing)	Families	20	19	18	94.7%	90.0%
Motta et al. 2018 (38)	Brazil	Individuals	N/A	21	19	90.5%	N/A
Neidhardt et al. 2008 (10)	Germany, The Netherlands, Denmark, and Switzerland	Individuals	90[b]	46	37	80.4%	41.1%
Oishi et al. 2014 (39)	Japan	Individuals	N/A	5	5	100%	N/A
Pelletier et al. 2007 (40)	France	Individuals	88	69	55	79.7%	62.5%
Perea-Romero et al. 2021 (41)	Spain	Families	N/A	91	68	74.7%	N/A
Pontikos et al. 2020 (42)	United Kingdom	Individuals	N/A	275	229	83.3%	N/A
Rodríguez-Muñoz et al.2020 (43)	Venezuela (Spanish origin)	Families	N/A	6	6	100%	N/A
Sharon and Banin 2015 (26)	Israel	Families	12	7	6	85.7%	50.0%
Sharon et al. 2003 (44)	Majority from United States and Canada	Individuals	187	90	80	88.9%	42.8%
Weisschuh et al. 2020 (23)	Germany (southwest)	Individuals	110	103	92	89.3%	83.6%
Yang et al. 2014 (45)	China (Beijing)	Families	7	Only *RPGR* screened	5	N/A	71.4%

N/A, not available; RP, retinitis pigmentosa; XLRP, X-linked retinitis pigmentosa. [a]54 cases from Germany and one case from each of the following countries: Croatia, Luxembourg, Switzerland, and Spain. [b]Number unclear in the manuscript.

Appendix 1. Search Strategy

Search 1: RP Prevalence

PubMed

Retinitis Pigmentosa OR Retinitis Pigmentosa[Title/Abstract] OR Tapetoretinal Degeneration*[Title/Abstract] OR Pigmentary Retinopath*[Title/Abstract]	AND	prevalence[MeSH] OR prevalen*[Title/Abstract] OR epidemiology [Subheading]	Limits: English, human

Embase

'pigmentary retinopathy':ab,ti OR 'pigmentary retinopathies':ab,ti OR 'tapetoretinal degeneration':ab,ti OR 'tapetoretinal degenerations':ab,ti OR 'retinitis pigmentosa':ab,ti OR 'retinitis pigmentosa'/de	AND	'epidemiology'/exp OR prevalen*:ab,ti OR 'prevalence'/exp	Limits: [english]/lim AND [embase]/lim AND ([article]/lim OR [article in press]/lim OR [review]/lim) AND [humans]/lim

Search 2: X-Linked Retinitis Pigmentosa (XLRP) Among Males With RP

PubMed

Retinitis Pigmentosa OR Retinitis Pigmentosa[Title/Abstract] OR Tapetoretinal Degeneration*[Title/Abstract] OR Pigmentary Retinopath*[Title/Abstract]	AND	X Chromosome[Mesh] OR X Chromosome[Title/Abstract] OR X-linked[Title/Abstract] OR XLRP[Title/Abstract]	AND	genetics [Subheading] OR DNA Mutational Analysis[Mesh:NoExp] OR Mutation[Mesh:NoExp] OR Mutation*[Title/Abstract]	AND	male*[Title/Abstract] OR men[Title/Abstract]	Limits: English Human

Embase

'pigmentary retinopathy':ab,ti OR 'pigmentary retinopathies':ab,ti OR 'tapetoretinal degeneration':ab,ti OR 'tapetoretinal degenerations':ab,ti OR 'retinitis pigmentosa':ab,ti OR 'retinitis pigmentosa'/de	AND	xlrp:ab,ti OR 'x chromosome':ab,ti OR 'x linked':ab,ti OR 'x chromosome linked disorder'/exp	AND	mutation*:ab,ti OR 'genetic analysis'/exp OR 'gene mutation'/exp	AND	men:ab,ti OR male*:ab,ti	Limits: [english]/lim AND [embase]/lim AND ([article]/lim OR [article in press]/lim OR [review]/lim) AND [humans]/lim

Search 3: RPGR Mutation Among Persons With XLRP

PubMed

Retinitis Pigmentosa OR Retinitis Pigmentosa[Title/Abstract] OR Tapetoretinal Degeneration*[Title/Abstract] OR Pigmentary Retinopath*[Title/Abstract]	AND	RPGR[Title/Abstract] OR DNA Mutational Analysis[Mesh:NoExp] OR Mutation[Mesh:NoExp] OR Mutation*[Title/Abstract]]	Limits: English Human

Embase

'pigmentary retinopathy':ab,ti OR 'pigmentary retinopathies':ab,ti OR 'tapetoretinal degeneration':ab,ti OR 'tapetoretinal degenerations':ab,ti OR 'retinitis pigmentosa':ab,ti OR 'retinitis pigmentosa'/de	AND	mutation*:ab,ti OR 'genetic analysis'/exp OR 'gene mutation'/exp OR rgpr:ti,ab	Limits: [english]/lim AND [embase]/lim AND ([article]/lim OR [article in press]/lim OR [review]/lim) AND [humans]/lim

Search 4: Prevalence and Genotype for Females

PubMed

Retinitis Pigmentosa OR Retinitis Pigmentosa[Title/Abstract] OR Tapetoretinal Degeneration*[Title/Abstract] OR Pigmentary Retinopath*[Title/Abstract]	AND	prevalence[MeSH] OR prevalen*[Title/Abstract] OR epidemiology [Subheading] OR genotype[MeSH] OR phenotype[MeSH] OR genotype*[Title/Abstract] OR phenotype*[Title/Abstract]	AND	female**[Title/Abstract] OR women*[Title/Abstract]	Limits: English, human

Embase

'pigmentary retinopathy':ab,ti OR 'pigmentary retinopathies':ab,ti OR 'tapetoretinal degeneration':ab,ti OR 'tapetoretinal degenerations':ab,ti OR 'retinitis pigmentosa':ab,ti OR 'retinitis pigmentosa'/de	AND	'epidemiology'/exp OR prevalen*:ab,ti OR 'prevalence'/exp OR 'genotype'/exp OR 'phenotype'/exp OR genotype*:ab,ti OR phenotype*:ab,ti	AND	Female*:ab,ti OR women:ab,ti	Limits: [english]/lim AND [embase]/lim AND ([article]/lim OR [article in press]/lim OR [review]/lim) AND [humans]/lim