Table S1. Molecular characterization of 1036 ar/sMD families over 30 years. Table S1A includes the study of 744 cases recruited since 1991 to 2017 in HUFJD. Table S1B includes the additional technologies used to characterize patients unsolved until 2017 (in blue). Table S1C includes the study of 292 cases recruited between January 2017 and October 2020 in HUFJD. #Gene panels has been previously described in Martin-Merida et al., 2019. $smMIPs technology has been previously reported in Khan et al., 2020. Cases in which c.4253+43G>A, c.4539+2064C>T, and c.5196+1137G>A variants have been sequenced are in orange. Cases without NGS studies are in red.

Table S1A

Technology	Initial cases (1991-2017)					
	solved 2017			unsolved 2017		
	STGD	CCRD	otherMD	STGD	CCRD	otherMD
Genotyping microarrays/Sanger sequencing	240	45	6	123	53	56
Gene panels (up to 82 genes)#	33	39	12	57	39	33
WES	x	5	x	1	2	x
Total	273	89	18	181	94	89

Table S1B

Technology	Re-studied cases (unsolved 1991-2017)					
	solved 2020			unsolved 2020		
	STGD	CCRD	otherMD	STGD	CCRD	otherMD
Gene panel with *ABCA4* variants reported by Braun et al., 2013.	18	1	x	1	1	1
Clinical exome (up to 229 genes)	32	23	28	4	17	31
smMIPs for entire *ABCA4* gene $ (in orange only prevalent DI *ABCA4* variants)	28	2	x	3 + 3	1	1
Clinical exome + smMIPs *ABCA4* (in orange only prevalent DI *ABCA4* variants)	x	x	x	18 + 4	2	x
WES	3	1	x	x	x	x
Total of re-studied cases	81	27	28	33	21	33
No more studies since 2017 (in red without NGS)	x	x	x	67 (53)	46 (23)	27 (15)

Table S1C

Technology	New cases (2017-2020)					
	solved 2020			unsolved 2020		
	STGD	CCRD	otherMD	STGD	CCRD	otherMD
Sequencing of the *ABCA4* gene (Asper Biogen)	18	x	x	x	x	4
Clinical exome (up to 229 genes)	66	24	44	12	14	80
Sequencing of the *ABCA4* gene (Asper Biogen) + smMIPs for entire *ABCA4* gene $	6	x	3	1	1	x
Clinical exome + smMIPs *ABCA4* (in orange only prevalent DI *ABCA4* variants)	x	x	x	10 + 2	1	5
Clinical exome + WES	x	x	x	1	x	x
Total	90	24	47	26	16	89

Abbreviations: STGD: Stargardt disease; CCRD: cone and cone-rod dystrophy; DI: deep intronic; otherMD; other maculopathy; smMIPs: single molecule molecular inversion probes; WES: whole exome sequencing; NGS: next generation sequencing technologies.