

Figure S1. Overview of characterization rates in patients re-studied using different NGS approaches, and the new cases studied by clinical exome and their classification by diagnosis clinical groups. Patients with * are patients recruited after 2017 re-studied by the sequencing of the entire *ABCA4* gene. Abbreviations: STGD: Stargardt; CCRD: cone and cone-rod dystrophy; otherMD: other maculopathies.

	Re-studied			New cases		
STGD diagnosis	114 17*	52%	<ul style="list-style-type: none"> Solved <i>ABCA4</i>: 66 + 6* (55%) Solved other genes: 15 (11%) Unsolved: 33 + 11* (34%) 	116 (40%)	<ul style="list-style-type: none"> Solved <i>ABCA4</i>: 87 (75%) Solved other genes: 3 (3%) Unsolved: 26 (22%) 	
CCRD diagnosis	48 2*	20%	<ul style="list-style-type: none"> Solved <i>ABCA4</i>: 15 (31%) Solved other genes: 12 (25%) Unsolved: 21 (44%) 	40 (14%)	<ul style="list-style-type: none"> Solved <i>ABCA4</i>: 12 (30%) Solved other genes: 12 (30%) Unsolved: 16 (40%) 	
otherMD diagnosis	61 8*	28%	<ul style="list-style-type: none"> Solved <i>ABCA4</i>: 4 + 3* (10%) Solved other genes: 24 (35%) Unsolved: 33 + 5* (55%) 	136 (46%)	<ul style="list-style-type: none"> Solved <i>ABCA4</i>: 25 (18%) Solved other genes: 22 (16%) Unsolved: 89 (66%) 	