

S1 Table. Current gene therapy trials for retinal dystrophies, including expected number of patients enrolled, timeframe and endpoints that assess efficacy.

NCT	Phase	Disease	Gene	Patients	Timeframe (months)	Endpoints for efficacy
01494805	I/II	AMD	<i>sFLT01</i>	40	36	BCVA, CNV lesion, foveal thickness
01301443	I	AMD	-	21	0,5	OCT
01024998	I	AMD	<i>sFLT01</i>	34	12	Decreased retinal thickness
02341807	I/II	CHM	<i>CHM</i>	10	24	Visual function
02077361	I	CHM	<i>CHM</i>	6	24	Visual field, visual function
01461213	I/II	CHM	<i>CHM</i>	14	6	Visual acuity
01496040	I/II	LCA	<i>RPE65</i>	9	12	ERG, patient questionnaire, visual acuity, color vision, pupillometry, microperimetry, dark adaptation
00999609	III	LCA	<i>RPE65</i>	24	12	Mobility testing
00821340	I	LCA	<i>RPE65</i>	10	36	Visual function
00749957	I/II	LCA	<i>RPE65</i>	12	24	Visual fields, visual acuity, ERG
00643747	I/II	LCA	<i>RPE65</i>	12	12	Visual function
00516477	I	LCA	<i>RPE65</i>	12	60	Visual function
00481546	I	LCA	<i>RPE65</i>	15	180	Visual function
01482195	I	LCA	<i>MERTK</i>	6	12	Visual function
01367444	I/II	STGD1	<i>ABCA4</i>	46	12	Delay in retinal degeneration by BCVA, microperimetry, full-field static/kinetic perimetry, OCT and FAF
02402660	II	STGD1	<i>ABCA4</i>	50	24	Combination of changes in atrophic lesion size, best-corrected visual acuity (BCVA), and ocular assessments
01278277	I/II	STGD1	<i>ABCA4</i>	30	6	Focal electroretinogram
01505062	I/II	USH1B	<i>MYO7A</i>	18	12	Delay in retinal degeneration
02317887	I/IIa	XLRS	<i>RS1</i>	24	18	Visual function, ERG, OCT, visual field

AMD = age-related macular degeneration, BCVA = best-corrected visual acuity, CHM = choroideremia, CNV = choroidal neovascularization, ERG = electroretinography, FAF = fundus autofluorescence, LCA = Leber's congenital amaurosis, NCT = national clinical trial, OCT = optical coherence tomography, STGD1 = Stargardt disease, USH1B = Usher Syndrome Type IB, XLRS = X-linked juvenile retinoschisis.