

Supplementary material for Wubben et al. 'Retinal detachment and infantile-onset glaucoma in Stickler syndrome associated with known and novel COL2A1 mutations.' *Ophthalmic Genetics*, 2018.

Supplemental Table 1. Genetic, systemic, and ophthalmic features in patients with Stickler syndrome.

Patient no.	Family no.	Age ^a	Sex	Mutation				Systemic features			Family History	Ocular Features (OD, OS)					Procedures (OD, OS)			Other remarks
				Exon/intron	Nucleotide change	Effect	Novel/Cited	Cleft palate	Micrognathia	Hearing loss		Visual acuity ^b	Refraction (D)	AL (mm)	RD	IOG	Prophy retinopathy	RD Sx	Glaucoma Sx	
1	1	2 years	M	35	c.2344_2355+5del GATGGTG GACGAGTA AG	USE	Novel	+	+	+	No	20/60, 20/60	-7.75, -7	NA, NA	-,-	-,-	-,-	-,-	-,-	Exophoria
2	2	4 mos.	M	51/51	c.3853G>T/ c.3641delC	p.Asp12 85Tyr/FS	Novel/ Cited (11)	+	+	+	Yes	F&F, F&F	-12.5, -13.5	28.7, 27.8	-,-	-,-	+, +	-,-	-,-	
3	3	2 mos.	F	IVS16	c.1023+6T>A	USE	Novel	+	-	-	No	20/30, 20/30	-5.3, -6	NA, NA	-,-	-,-	+, +	-,-	-,-	
4	4	10 mos.	F	23	c.1475G>T	p.Gly492 Val	Cited (14)	+	+	+	Yes	20/125, 20/30	-5.3, -0.5	25.4, 23.0	-,-	-,-	+, +	-,-	-,-	Amblyopia
5	4	29 years	F	23	c.1475G>T	p.Gly492 Val	Cited (14)	+	-	+	Yes	20/20, 20/25	-1.0, -1.3	NA, NA	-,-	-,-	-,-	-,-	-,-	
6	5	1 year	M	40	c.2659C>T	p.Arg887 X	Cited (15)	-	-	+	Yes	20/70, 20/40	-10.6, -3.3	NA, NA	-,-	-,-	+, +	-,-	-,-	Amblyopia, Perthes disease
7	5	22 years	M	NA	NA	NA	NA	NA	NA	NA	Yes	20/40, 20/25	NA, NA	NA, NA	-,-	-,-	+, +	-,-	-,-	Glaucoma suspect
8	6	26 years	F	41	c.2710C>T	p.Arg904 Cys	Cited (16)	-	+	+	Yes	20/20, NLP	-19.3, NA	NA, NA	-,-	-,-	+, +	-,-	-,-	Phthisis OS
9	6	3 wks	M	NA	NA	NA	NA	-	-	+	Yes	NLP, 20/70	-22.8, -20.5	29.1, 29.6	+, +	+, +	+, +	PPV, PPV,-	trabeculotomy/ trab /trab with MMC/BV350, trabeculotomy/ trab/trab with MMC/BV350/ CPCx3	Phthisis OD, CE/IOL OS
10	6	3 mos.	M	NA	NA	NA	NA	+	-	+	Yes	NLP, 20/100	-19, -16	29.3, 30.6	+, +	+, +	-,-	PPV, -	Trabeculotomyx 4/trab/ trab with MMC/BV350, trabeculotomyx 4/trab/ trab with MMC/trab/ BV350	Endophthalmitis OD, Phthisis OD, CE/IOL and PKP OS
11	7	1 year	M	26	c.1693C>T	p.Arg565 Cys	Cited (16)	-	+	+	No	20/60, 20/40	-15.4, -13.4	28.1, 27.7	-,-	-,-	+, +	-,-	-,-	Amblyopia, Scimitar syndrome
12	8	9 years	M	9	c.625C>T	p.Arg209 X	Cited (11)	+	+	+	Yes	NLP, 20/32	NA, -8.8	31.8, 27.4	+, +	+, +	-,-	PPVx 3, -	BV250	
13	9	48 years	F	NA	NA	NA	NA	NA	NA	NA	Yes	Prosthetic, 20/25	NA, -10.5	NA, NA	+, +	-,-	-,-	SB, PPVx 5, -	-,-	Prosthetic OD, CE/IOL OS
14	10	32 years	M	NA	NA	NA	NA	NA	NA	NA	Yes	20/25, 20/50	-7.0, -9.0	NA, NA	-,-	-,-	-,-	-,-	-,-	
15	11	18 years	M	NA	NA	NA	NA	+	+	-	Yes	20/20, 20/25	-6.5, -10.0	NA, NA	-,-	-,-	+, +	-,-	-,-	Focal cataracts

^aAge at initial presentation

^bVisual acuity at latest follow up visit

M, male; F, female; USE, undetermined splicing effect; FS, frameshift; IVS, intervening sequence; NA, not available; wks, weeks; mos., months; D, diopters; mm, millimeters; F&F, fix and follow; NLP, no light perception; PPV, pars plana vitrectomy; trab, trabeculectomy; MMC, mitomycin C; BV, baerveldt; CPC, cyclophotocoagulation; PKP, penetrating keratoplasty; CE/IOL, cataract extraction and intraocular lens placement; SB, scleral buckle; AL, axial length; RD, retinal detachment; IOG, infantile-onset glaucoma; Sx, surgery