

## SUPPLEMENTAL MATERIAL

Ward et al., <http://www.jem.org/cgi/content/full/jem.20140214/DC1>**Clinical characteristics of human subjects who underwent retinal imaging**

	Matched controls	All <i>GRN</i> mutation carriers	p value (ctrl vs <i>GRN</i> mt)
n (patients): RNFL	24	12	
n (patients): MacV	24	11	
n (eyes): RNFL	48	24	
n (eyes): MacV	48	22	
age yr (S.D.)	57.3 (11.34)	55.5 (10.4)	
male sex no. (%)	6 (25)	4 (33)	
CDR (S.D.)	0 (0)	0.5 (0.8)	
mean RNFL thickness $\mu\text{m}$ (S.D.)	98.0 (7.6)	90.4 (7.7)	p = .001
mean MacV $\text{mm}^3$ (S.D.)	3.13 (0.14)	3.00 (0.13)	p = .004
mean GCC volume $\text{mm}^3$ (S.D.)	0.81 (0.07)	0.75 (0.07)	p = .002
mean INL volume $\text{mm}^3$ (S.D.)	0.33 (0.03)	0.31 (0.03)	p = .011
mean ONL volume $\text{mm}^3$ (S.D.)	0.69 (0.07)	0.66 (0.06)	p = .077

	Matched controls	Pre-symptomatic <i>GRN</i> mutation carriers (CDR = 0)	p value (ctrl vs <i>GRN</i> mt)
n (patients): RNFL	14	7	
n (patients): MacV	14	7	
n (eyes): RNFL	28	14	
n (eyes): MacV	28	14	
age yr (S.D.)	50.6 (10.7)	49.9 (9.1)	
male sex no. (%)	3 (22)	3 (43)	
CDR (S.D.)	0	0	
mean RNFL thickness $\mu\text{m}$ (S.D.)	100.1 (7.2)	90.6 (9.2)	p = .003
mean MacV $\text{mm}^3$ (S.D.)	3.19 (.12)	3.05 (0.11)	p = .016
mean GCC volume $\text{mm}^3$ (S.D.)	0.84 (.04)	0.76 (.06)	p < .001
mean INL volume $\text{mm}^3$ (S.D.)	0.33 (.03)	0.32 (0.03)	p = .163
mean ONL volume $\text{mm}^3$ (S.D.)	0.71 (0.06)	0.68 (0.04)	p = .174

	Matched controls	Symptomatic <i>GRN</i> mutation carriers (CDR > 0)	p value (ctrl vs <i>GRN</i> mt)
n (patients): RNFL	10	5	
n (patients): MacV	10	4	
n (eyes): RNFL	20	10	
n (eyes): MacV	20	8	
age yr (S.D.)	63.4 (6.7)	63.4 (6.1)	
male sex no. (%)	3 (30)	1 (20)	
CDR (S.D.)	0	1.2 (0.8)	
mean RNFL thickness $\mu\text{m}$ (S.D.)	95.0 (7.3)	90.3 (5.3)	p = 0.149
mean MacV $\text{mm}^3$ (S.D.)	3.05 (0.12)	2.91 (0.12)	p = 0.024
mean GCC volume $\text{mm}^3$ (S.D.)	0.77 (0.09)	0.72 (0.09)	p = 0.219
mean INL volume $\text{mm}^3$ (S.D.)	0.33 (0.03)	0.30 (0.01)	p = 0.011
mean ONL volume $\text{mm}^3$ (S.D.)	0.66 (0.08)	0.61 (0.07)	p = 0.254

**Figure S1. Clinical characteristics of human subjects who underwent retinal imaging.** Quantification of OCT images was performed by automated software algorithms (Heidelberg Engineering, Heidelberg, Germany). P-values were determined via mixed effects linear regression analysis.