

Supplementary Materials

Risk of Optic Pathway Glioma in Neurofibromatosis Type 1: No Evidence of Genotype–Phenotype Correlations in A Large Independent Cohort

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Table S1. Mutations in *NF1* gene tertiles and the risk of developing OPG in the combined cohort of our and Anastasaki's patients.

Tertile	OPG <i>n</i> (%) <i>n</i> = 259	Non-OPG <i>n</i> (%) <i>n</i> = 431	<i>p</i> -Value *	OR (95% CI)	<i>p</i> -Value **	Total Number <i>n</i> = 690
5' Tertile	131 (50.6)	187 (43.4)	0.066	1.33 (0.98- 1.81)	0.067	318
Middle tertile	82 (31.7)	154 (35.7)	0.27	0.83 (0.60-1.15)	0.27	236
3' Tertile	46 (17.8)	90 (20.9)	0.31	0.81 (0.55-1.2)	0.53	136

Differences in the frequency of mutations in the different tertiles of the *NF1* gene between the OPG and the non-OPG group. *Chi-squared test; ** logistic regression.

Table S2. Mutations in different *NF1* gene regions and the risk of developing OPG in the combined cohort of our and Anastasaki's patients.

Regions	OPG <i>n</i> (%) <i>n</i> = 259	Non-OPG <i>n</i> (%) <i>n</i> = 431	<i>p</i> -Value *	OR (95% CI)	<i>p</i> -Value **	Total Number <i>n</i> = 690
CSRD	49 (18.9)	60 (13.9)	0.81	1.44 (0.95-2.1)	0.82	109
TBD	5 (1.9)	20 (4.6)	0.65	0.40 (0.15 -1)	0.74	25
GRD	41 (15.8)	63 (14.6)	0.66	1.09 (0.71- 1.68)	0.66	104
Sec14-PH	22 (8.5)	24 (5.6)	0.13	1.57 (0.86 -2.86)	0.13	46
HLR	36 (13.9)	78 (18.1)	0.15	0.73 (0.47-1.1)	0.15	114
CTD	25 (9.7)	43(10)	0.89	0.96 (0.57 - 1.6)	0.89	68
NLS	0	1 (0.2)	1	1 (0.99 - 1)	1	1
SBR	0	2 (0.5)	0.53	1 (0.99 - 1.01)	0.99	2
Others	99 (38.2)	169 (39.2)	0.79	0.95 (0.69- 1.31)	0.79	268

Differences in the frequency of mutations in the different domains of the *NF1* gene between the OPG and the non-OPG group. * Chi-squared test; ** logistic regression.



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