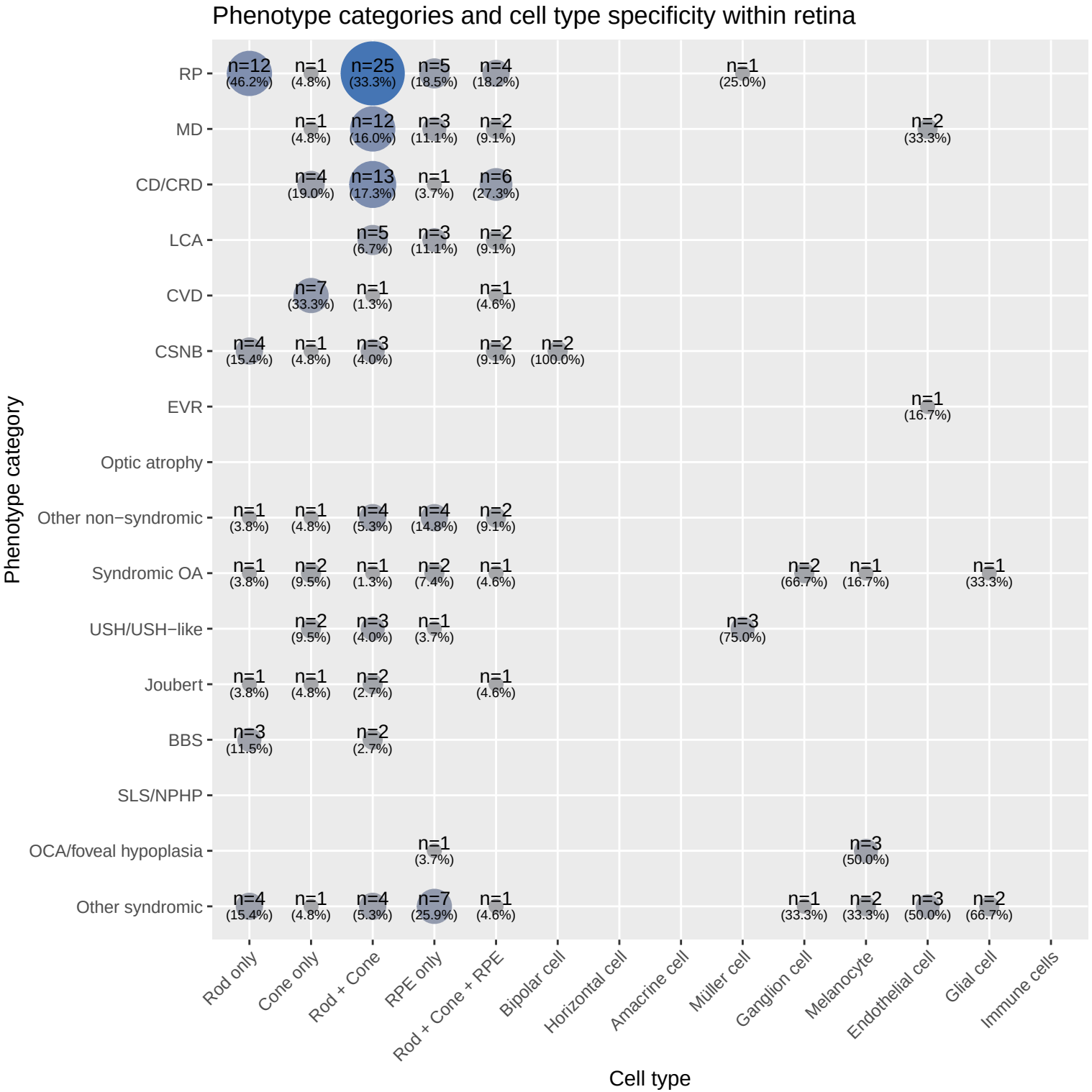
Phenotype categories and cell type specificity within retina
Phenotype category
RP
MD
CD/CRD
LCA
CVD
CSNB
EVR
Optic atrophy
Other non−syndromic
Syndromic OA
USH/USH−like
Joubert
BBS
SLS/NPHP
OCA/foveal hypoplasia
Other syndromic
n=12 (46.2%)
n=1 (4.8%)
n=25 (33.3%)
n=5 (18.5%)
n=4 (18.2%)
n=1 (25.0%)
n=1 (4.8%)
n=12 (16.0%)
n=3 (11.1%)
n=2 (9.1%)
n=2 (33.3%)
n=4 (19.0%)
n=13 (17.3%)
n=1 (3.7%)
n=6 (27.3%)
n=5 (6.7%)
n=3 (11.1%)
n=2 (9.1%)
n=7 (33.3%)
n=1 (1.3%)
n=1 (4.6%)
n=4 (15.4%)
n=1 (4.8%)
n=3 (4.0%)
n=2 (9.1%)
n=2 (100.0%)
n=1 (16.7%)
n=1 (3.8%)
n=1 (4.8%)
n=4 (5.3%)
n=4 (14.8%)
n=2 (9.1%)
n=1 (3.8%)
n=2 (9.5%)
n=1 (1.3%)
n=2 (7.4%)
n=1 (4.6%)
n=2 (66.7%)
n=1 (16.7%)
n=1 (33.3%)
n=2 (9.5%)
n=3 (4.0%)
n=1 (3.7%)
n=3 (75.0%)
n=1 (3.8%)
n=1 (4.8%)
n=2 (2.7%)
n=1 (4.6%)
n=3 (11.5%)
n=2 (2.7%)
n=1 (3.7%)
n=3 (50.0%)
n=4 (15.4%)
n=1 (4.8%)
n=4 (5.3%)
n=7 (25.9%)
n=1 (4.6%)
n=1 (33.3%)
n=2 (33.3%)
n=3 (50.0%)
n=2 (66.7%)
Rod only
Cone only
Rod + Cone
RPE only
Rod + Cone + RPE
Bipolar cell
Horizontal cell
Amacrine cell
Müller cell
Ganglion cell
Melanocyte
Endothelial cell
Glial cell
Immune cells
Cell type