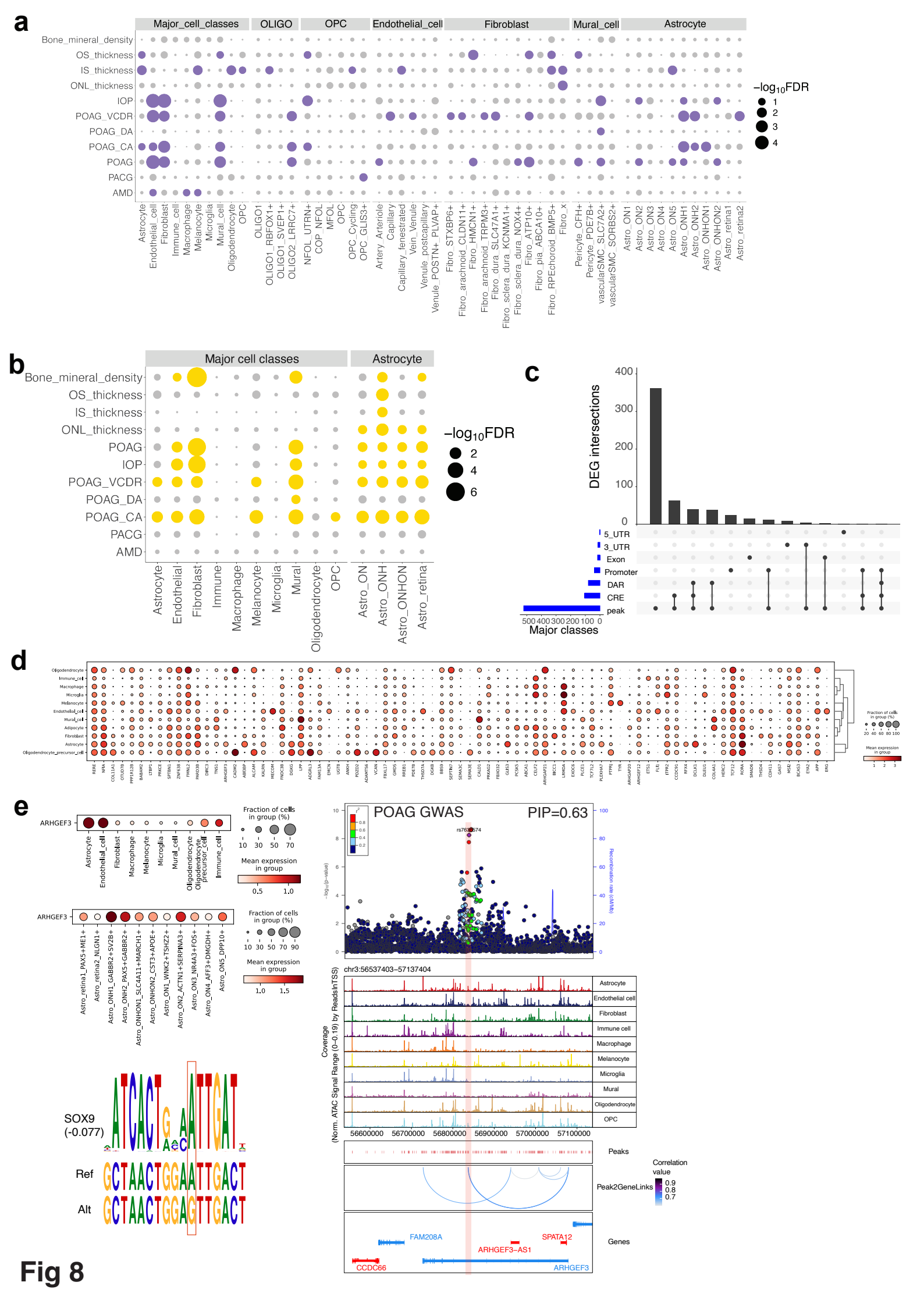
a
Major_cell_classes
OLIGO
OPC
Endothelial_cell
Fibroblast
Mural_cell
Astrocyte
Bone_mineral_density
OS_thickness
IS_thickness
ONL_thickness
IOP
POAG_VCDR
POAG_DA
POAG_CA
POAG
PACG
AMD
−log10FDR
1
2
3
4
Astrocyte
Endothelial_cell
Fibroblast
Immune_cell
Macrophage
Melanocyte
Microglia
Mural_cell
Oligodendrocyte
OPC
OLIGO1
OLIGO1_RBFOX1+
OLIGO1_SVEP1+
OLIGO2_LRRC7+
NFOL_UTRN+
COP_NFOL
MFOL
OPC
OPC_Cycling
OPC_GLIS3+
Artery_Arteriole
Capillary
Capillary_fenestrated
Vein_Venule
Venule_postcapillary
Venule_POSTN+_PLVAP+
Fibro_STXBP6+
Fibro_arachnoid_CLDN11+
Fibro_HMCN1+
Fibro_arachnoid_TRPM3+
Fibro_dura_SLC47A1+
Fibro_sclera_dura_KCNMA1+
Fibro_sclera_dura_NOX4+
Fibro_ATP10+
Fibro_pia_ABCA10+
Fibro_RPEchoroid_BMP5+
Fibro_x
Pericyte_CFH+
Pericyte_PDE7B+
vascularSMC_SLC7A2+
vascularSMC_SORBS2+
Astro_ON1
Astro_ON2
Astro_ON3
Astro_ON4
Astro_ON5
Astro_ONH1
Astro_ONH2
Astro_ONHON1
Astro_ONHON2
Astro_retina1
Astro_retina2
b
Major cell classes
Astrocyte
Bone_mineral_density
OS_thickness
IS_thickness
ONL_thickness
POAG
IOP
POAG_VCDR
POAG_DA
POAG_CA
PACG
AMD
−log10FDR
2
4
6
Astrocyte
Endothelial
Fibroblast
Immune
Macrophage
Melanocyte
Microglia
Mural
Oligodendrocyte
OPC
Astro_ON
Astro_ONH
Astro_ONHON
Astro_retina
c
DEG intersections
400
300
200
100
0
5_UTR
3_UTR
Exon
Promoter
DAR
CRE
peak
500 400 300 200 100 0
Major classes
d
Oligodendrocyte
Immune_cell
Macrophage
Microglia
Melanocyte
Endothelial_cell
Mural_cell
Adipocyte
Fibroblast
Astrocyte
Oligodendrocyte_precursor_cell
Fraction of cells in group (%)
20 40 60 80 100
Mean expression in group
1 2 3
e
ARHGEF3
Astrocyte
Endothelial_cell
Fibroblast
Macrophage
Melanocyte
Microglia
Mural_cell
Oligodendrocyte
Oligodendrocyte precursor_cell
Immune_cell
Fraction of cells in group (%)
10 30 50 70
Mean expression in group
0.5 1.0
ARHGEF3
Astro_retina1_PAX5+ME1+
Astro_retina2_NLGN1+
Astro_ONH1_GABBR2+SV2B+
Astro_ONH2_PAX5+GABBR2+
Astro_ONHON1_SLC4A11+MARCH1+
Astro_ONHON2_CST3+APOE+
Astro_ON1_WNK2+TSHZ2+
Astro_ON2_ACTN1+SERPINA3+
Astro_ON3_NR4A3+FOS+
Astro_ON4_AFF3+DMGDH+
Astro_ON5_DPP10+
Fraction of cells in group (%)
10 30 50 70 90
Mean expression in group
1.0 1.5
SOX9
(-0.077)
Ref
Alt
POAG GWAS
PIP=0.63
rs7632574
−log10(p-value)
Recombination rate (cM/Mb)
chr3:56537403−57137404
Coverage
(Norm. ATAC Signal Range (0−0.19) by ReadsInTSS)
Astrocyte
Endothelial cell
Fibroblast
Immune cell
Macrophage
Melanocyte
Microglia
Mural
Oligodendrocyte
OPC
56600000 56700000 56800000 56900000 57000000 57100000
Peaks
Correlation value
Peak2GeneLinks
0.9
0.8
0.7
Genes
FAM208A
SPATA12
ARHGEF3−AS1
CCDC66
ARHGEF3

Fig 8

Figure 8. GWAS cell type enrichment and fine-mapping
a, Cell class enrichment of GWAS loci based on snRNA-seq gene expression with MAGMA. Celltyping. Rows represent enriched GWAS traits, and columns represent optic nerve cell classes and cell types. The dot size indicates −log10P. Significant results (FDR < 0.1) are highlighted in purple, while non-significant results (FDR ≥ 0.1) are shown in grey. **b,** Cell class enrichment of GWAS loci based on snATAC-seq OCRs across major cell classes with LDSC. Rows represent enriched GWAS traits, and columns represent optic nerve cell classes. The dot size indicates $-\log_{10}$FDR. Significant results (FDR < 0.1) are highlighted in yellow, while non-significant results (FDR ≥ 0.1) are shown in grey. **c**, Categorization of fine-mapped GWAS variants according to their genomic locations. Categories include peak (namely, OCRs), linked CREs, DARs, promoter, exon, 5′ UTR and 3′ UTR. **d**, Dot plot showing the gene expression of prioritized GWAS genes. **e**, A fine-mapped POAG GWAS locus, rs7636574, in the *ARHGEF3* region.