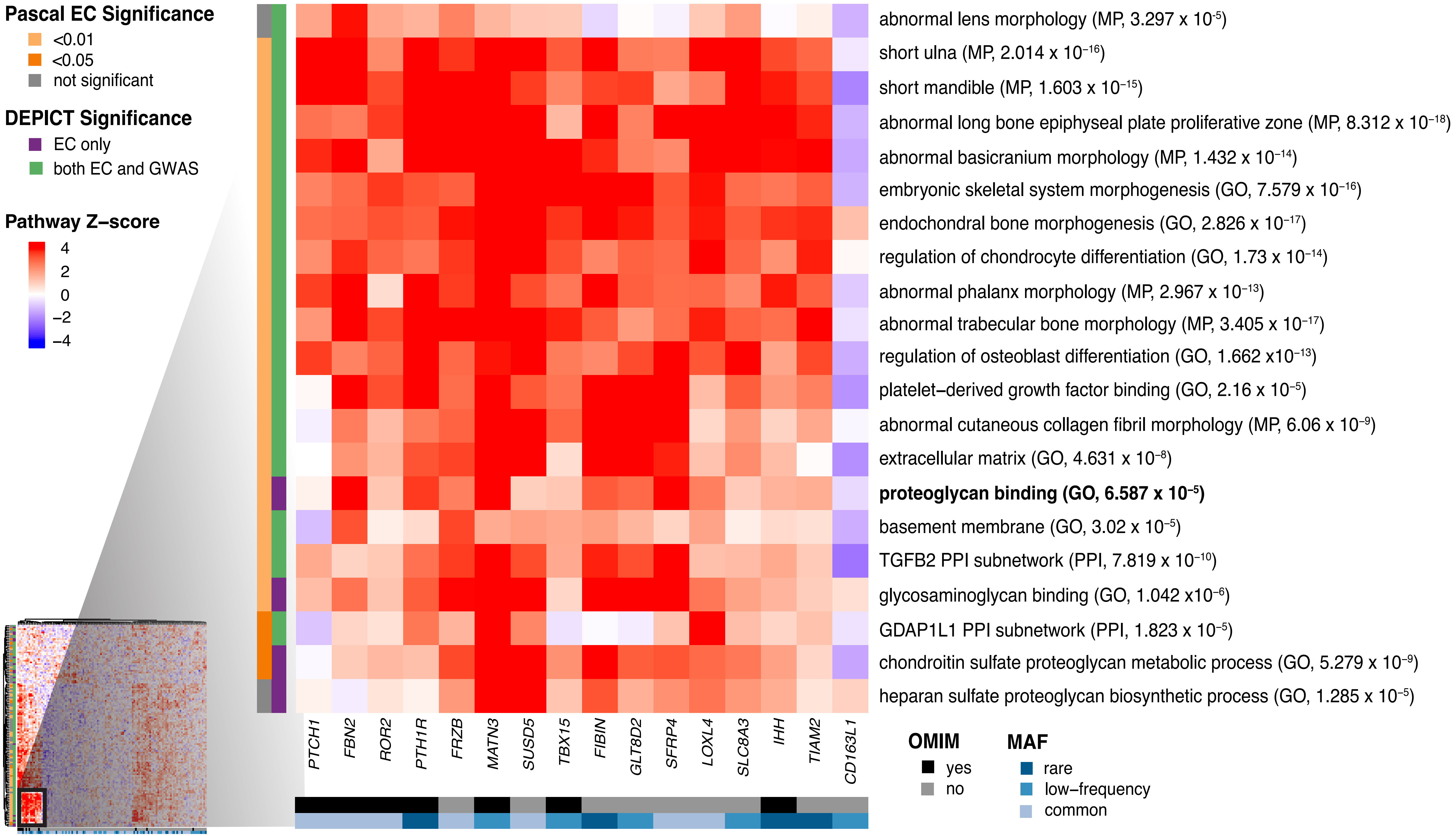
Pascal EC Significance
<0.01
<0.05
not significant
DEPICT Significance
EC only
both EC and GWAS
Pathway Z–score
4
2
0
−2
−4
abnormal lens morphology (MP, 3.297 x 10^{-5})
short ulna (MP, 2.014 x 10^{-16})
short mandible (MP, 1.603 x 10^{-15})
abnormal long bone epiphyseal plate proliferative zone (MP, 8.312 x 10^{-18})
abnormal basicranium morphology (MP, 1.432 x 10^{-14})
embryonic skeletal system morphogenesis (GO, 7.579 x 10^{-16})
endochondral bone morphogenesis (GO, 2.826 x 10^{-17})
regulation of chondrocyte differentiation (GO, 1.73 x 10^{-14})
abnormal phalanx morphology (MP, 2.967 x 10^{-13})
abnormal trabecular bone morphology (MP, 3.405 x 10^{-17})
regulation of osteoblast differentiation (GO, 1.662 x10^{-13})
platelet–derived growth factor binding (GO, 2.16 x 10^{-5})
abnormal cutaneous collagen fibril morphology (MP, 6.06 x 10^{-9})
extracellular matrix (GO, 4.631 x 10^{-8})
proteoglycan binding (GO, 6.587 x 10^{-5})
basement membrane (GO, 3.02 x 10^{-5})
TGFB2 PPI subnetwork (PPI, 7.819 x 10^{-10})
glycosaminoglycan binding (GO, 1.042 x10^{-6})
GDAP1L1 PPI subnetwork (PPI, 1.823 x 10^{-5})
chondroitin sulfate proteoglycan metabolic process (GO, 5.279 x 10^{-9})
heparan sulfate proteoglycan biosynthetic process (GO, 1.285 x 10^{-5})
PTCH1
FBN2
ROR2
PTH1R
FRZB
MATN3
SUSD5
TBX15
FIBIN
GLT8D2
SFRP4
LOXL4
SLC8A3
IHH
TIAM2
CD163L1
OMIM
yes
no
MAF
rare
low–frequency
common