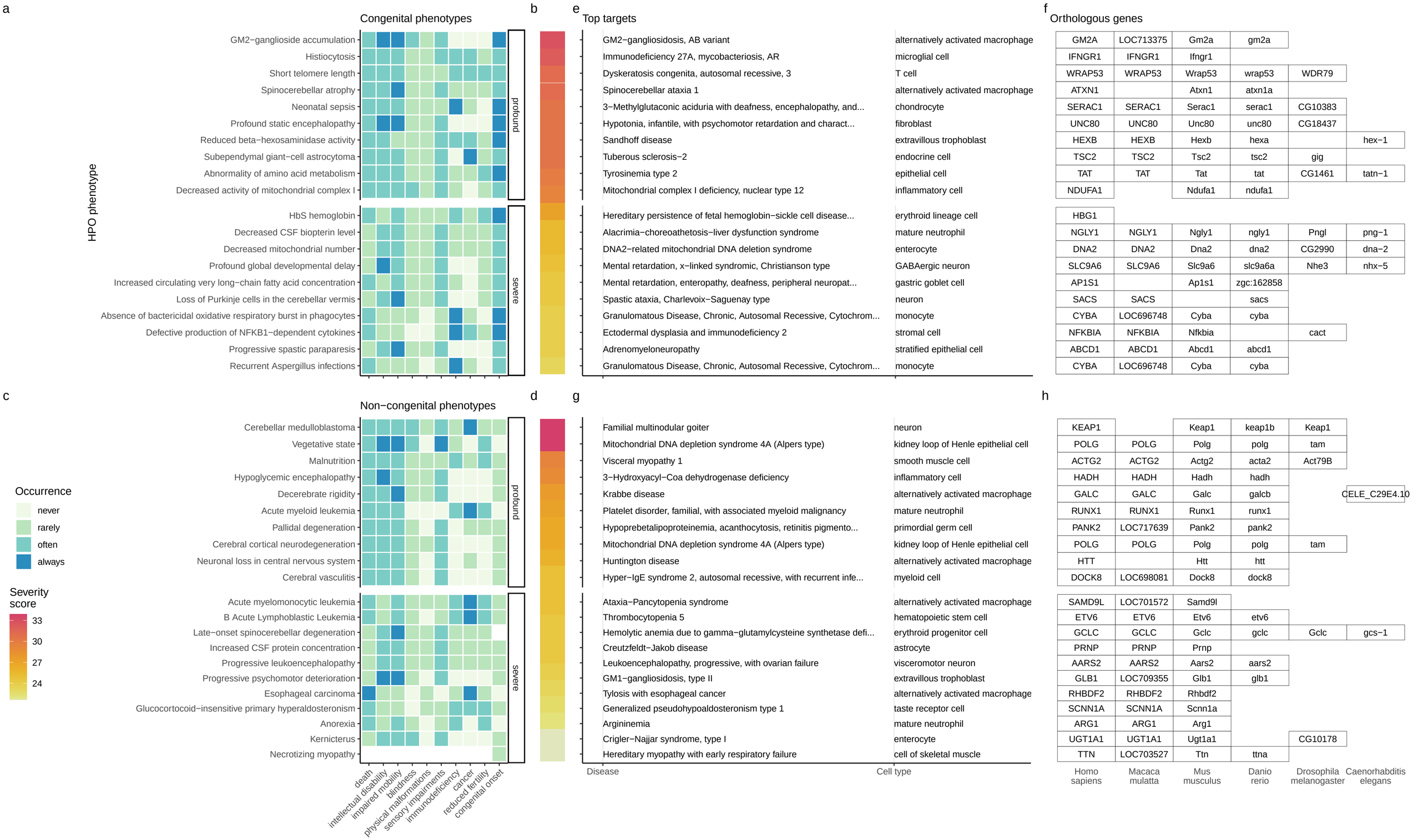
a
Congenital phenotypes
HPO phenotype
GM2−ganglioside accumulation
Histiocytosis
Short telomere length
Spinocerebellar atrophy
Neonatal sepsis
Profound static encephalopathy
Reduced beta−hexosaminidase activity
Subependymal giant−cell astrocytoma
Abnormality of amino acid metabolism
Decreased activity of mitochondrial complex I
HbS hemoglobin
Decreased CSF biopterin level
Decreased mitochondrial number
Profound global developmental delay
Increased circulating very long−chain fatty acid concentration
Loss of Purkinje cells in the cerebellar vermis
Absence of bactericidal oxidative respiratory burst in phagocytes
Defective production of NFKB1−dependent cytokines
Progressive spastic paraparesis
Recurrent Aspergillus infections
profound
severe
b
e
Top targets
Disease
Cell type
GM2−gangliosidosis, AB variant
alternatively activated macrophage
Immunodeficiency 27A, mycobacteriosis, AR
microglial cell
Dyskeratosis congenita, autosomal recessive, 3
T cell
Spinocerebellar ataxia 1
alternatively activated macrophage
3−Methylglutaconic aciduria with deafness, encephalopathy, and...
chondrocyte
Hypotonia, infantile, with psychomotor retardation and charact...
fibroblast
Sandhoff disease
extravillous trophoblast
Tuberous sclerosis−2
endocrine cell
Tyrosinemia type 2
epithelial cell
Mitochondrial complex I deficiency, nuclear type 12
inflammatory cell
Hereditary persistence of fetal hemoglobin−sickle cell disease...
erythroid lineage cell
Alacrimia−choreoathetosis−liver dysfunction syndrome
mature neutrophil
DNA2−related mitochondrial DNA deletion syndrome
enterocyte
Mental retardation, x−linked syndromic, Christianson type
GABAergic neuron
Mental retardation, enteropathy, deafness, peripheral neuropat...
gastric goblet cell
Spastic ataxia, Charlevoix−Saguenay type
neuron
Granulomatous Disease, Chronic, Autosomal Recessive, Cytochrom...
monocyte
Ectodermal dysplasia and immunodeficiency 2
stromal cell
Adrenomyeloneuropathy
stratified epithelial cell
Granulomatous Disease, Chronic, Autosomal Recessive, Cytochrom...
monocyte
f
Orthologous genes
Homo sapiens
Macaca mulatta
Mus musculus
Danio rerio
Drosophila melanogaster
Caenorhabditis elegans
GM2A | LOC713375 | Gm2a | gm2a
IFNGR1 | IFNGR1 | Ifngr1
WRAP53 | WRAP53 | Wrap53 | wrap53 | WDR79
ATXN1 | Atxn1 | atxn1a
SERAC1 | SERAC1 | Serac1 | serac1 | CG10383
UNC80 | UNC80 | Unc80 | unc80 | CG18437
HEXB | HEXB | Hexb | hexa | hex−1
TSC2 | TSC2 | Tsc2 | tsc2 | gig
TAT | TAT | Tat | tat | CG1461 | tatn−1
NDUFA1 | Ndufa1 | ndufa1
HBG1
NGLY1 | NGLY1 | Ngly1 | ngly1 | Pngl | png−1
DNA2 | DNA2 | Dna2 | dna2 | CG2990 | dna−2
SLC9A6 | SLC9A6 | Slc9a6 | slc9a6a | Nhe3 | nhx−5
AP1S1 | Ap1s1 | zgc:162858
SACS | SACS | sacs
CYBA | LOC696748 | Cyba | cyba
NFKBIA | NFKBIA | Nfkbia | cact
ABCD1 | ABCD1 | Abcd1 | abcd1
CYBA | LOC696748 | Cyba | cyba
c
Non−congenital phenotypes
Occurrence
never
rarely
often
always
Severity score
33
30
27
24
Cerebellar medulloblastoma
Vegetative state
Malnutrition
Hypoglycemic encephalopathy
Decerebrate rigidity
Acute myeloid leukemia
Pallidal degeneration
Cerebral cortical neurodegeneration
Neuronal loss in central nervous system
Cerebral vasculitis
Acute myelomonocytic leukemia
B Acute Lymphoblastic Leukemia
Late−onset spinocerebellar degeneration
Increased CSF protein concentration
Progressive leukoencephalopathy
Progressive psychomotor deterioration
Esophageal carcinoma
Glucocortocoid−insensitive primary hyperaldosteronism
Anorexia
Kernicterus
Necrotizing myopathy
profound
severe
death
intellectual disability
impaired mobility
blindness
physical malformations
sensory impairments
immunodeficiency
cancer
reduced fertility
congenital onset
d
g
Familial multinodular goiter
neuron
Mitochondrial DNA depletion syndrome 4A (Alpers type)
kidney loop of Henle epithelial cell
Visceral myopathy 1
smooth muscle cell
3−Hydroxyacyl−Coa dehydrogenase deficiency
inflammatory cell
Krabbe disease
alternatively activated macrophage
Platelet disorder, familial, with associated myeloid malignancy
mature neutrophil
Hypoprebetalipoproteinemia, acanthocytosis, retinitis pigmento...
primordial germ cell
Mitochondrial DNA depletion syndrome 4A (Alpers type)
kidney loop of Henle epithelial cell
Huntington disease
alternatively activated macrophage
Hyper−IgE syndrome 2, autosomal recessive, with recurrent infe...
myeloid cell
Ataxia−Pancytopenia syndrome
alternatively activated macrophage
Thrombocytopenia 5
hematopoietic stem cell
Hemolytic anemia due to gamma−glutamylcysteine synthetase defi...
erythroid progenitor cell
Creutzfeldt−Jakob disease
astrocyte
Leukoencephalopathy, progressive, with ovarian failure
visceromotor neuron
GM1−gangliosidosis, type II
extravillous trophoblast
Tylosis with esophageal cancer
alternatively activated macrophage
Generalized pseudohypoaldosteronism type 1
taste receptor cell
Argininemia
mature neutrophil
Crigler−Najjar syndrome, type I
enterocyte
Hereditary myopathy with early respiratory failure
cell of skeletal muscle
h
KEAP1 | Keap1 | keap1b | Keap1
POLG | POLG | Polg | polg | tam
ACTG2 | ACTG2 | Actg2 | acta2 | Act79B
HADH | HADH | Hadh | hadh
GALC | GALC | Galc | galcb | CELE_C29E4.10
RUNX1 | RUNX1 | Runx1 | runx1
PANK2 | LOC717639 | Pank2 | pank2
POLG | POLG | Polg | polg | tam
HTT | Htt | htt
DOCK8 | LOC698081 | Dock8 | dock8
SAMD9L | LOC701572 | Samd9l
ETV6 | ETV6 | Etv6 | etv6
GCLC | GCLC | Gclc | gclc | Gclc | gcs−1
PRNP | PRNP | Prnp
AARS2 | AARS2 | Aars2 | aars2
GLB1 | LOC709355 | Glb1 | glb1
RHBDF2 | RHBDF2 | Rhbdf2
SCNN1A | SCNN1A | Scnn1a
ARG1 | ARG1 | Arg1
UGT1A1 | UGT1A1 | Ugt1a1 | CG10178
TTN | LOC703527 | Ttn | ttna