

# Phenotypes

Phenotypes

AllNone

☒autism

☒congenital heart disease

☒epilepsy

☒intellectual disability

☒schizophrenia

☒unaffected

☐

☐

☐

☐

☐

☐

Present in Child and Present in Parent

Present in Child

AllNone

☒ affected only

☒ unaffected only

☒ affected and unaffected

☒ neither

Default selection should be 'affected only' and 'affected and unaffected'

'neither' option should appear only if the dataset in use has transmitted variants

Present in Parent

AllNone

☒ mother only

☒ father only

☒ mother and father

☒ neither

Default selection should be 'neither'

'neither' option should appear only if the dataset in use has denovo variants

Effect Types

Effect Types

AllNoneLGDsnonsynonymousUTRs

Coding

☒ nonsense

☒ frame-shift

☒ splice-site

☒ missense

☒ non-frame-shift

☒ no start

☒ no end

☒ synonymous

Noncoding

☒ non coding

☒ intron

☒ intergenic

☒ 3' UTR

☒ 5' UTR

CNV

☒ CNV+

☒ CNV-

LGDs button selects 'nonsense', 'frame-shift' and 'splice-site'

nonsynonymous button selects all effects in 'Coding' column except 'synonymous'

UTRs button selects '3'UTR' and '5'UTR'

CNV should appear only if the dataset has CNV variants

Variant and Study types, Gender

Child Gender

AllNone

☒ ☐

☒ ☐

Variant Types

AllNone

☒ sub

☒ ins

☒ del

☒ CNV

Study Types

AllNone

☒ WE

☒ TG

☒ WG