

**New Diagnoses
(Count)**

Full Cohort
(248)

NDD Sub-cohort
(91)

Cardiac Sub-cohort
(54)

Renal Sub-cohort
(43)

Gene-disease
Associations

Variant
Reclassifications

Missed in
Manual Analysis

CNV/SV




Total Cohort Size
New Diagnosis %



N=4738
5.2%





N=1665
5.4%

N=997
5.4%

N=695
6.1%

 New MOI for known gene and phenotype
 New gene-disease association
 New phenotype for known gene

 New ClinVar entries
 New classification rules

 Human error
 Limited Panel
 Non-coding variant
 QC Fail

 CNV
 SV

