THIRD INTERNATIONAL CONFERENCE ON RARE AND UNDIAGNOSED DISEASES

Update from
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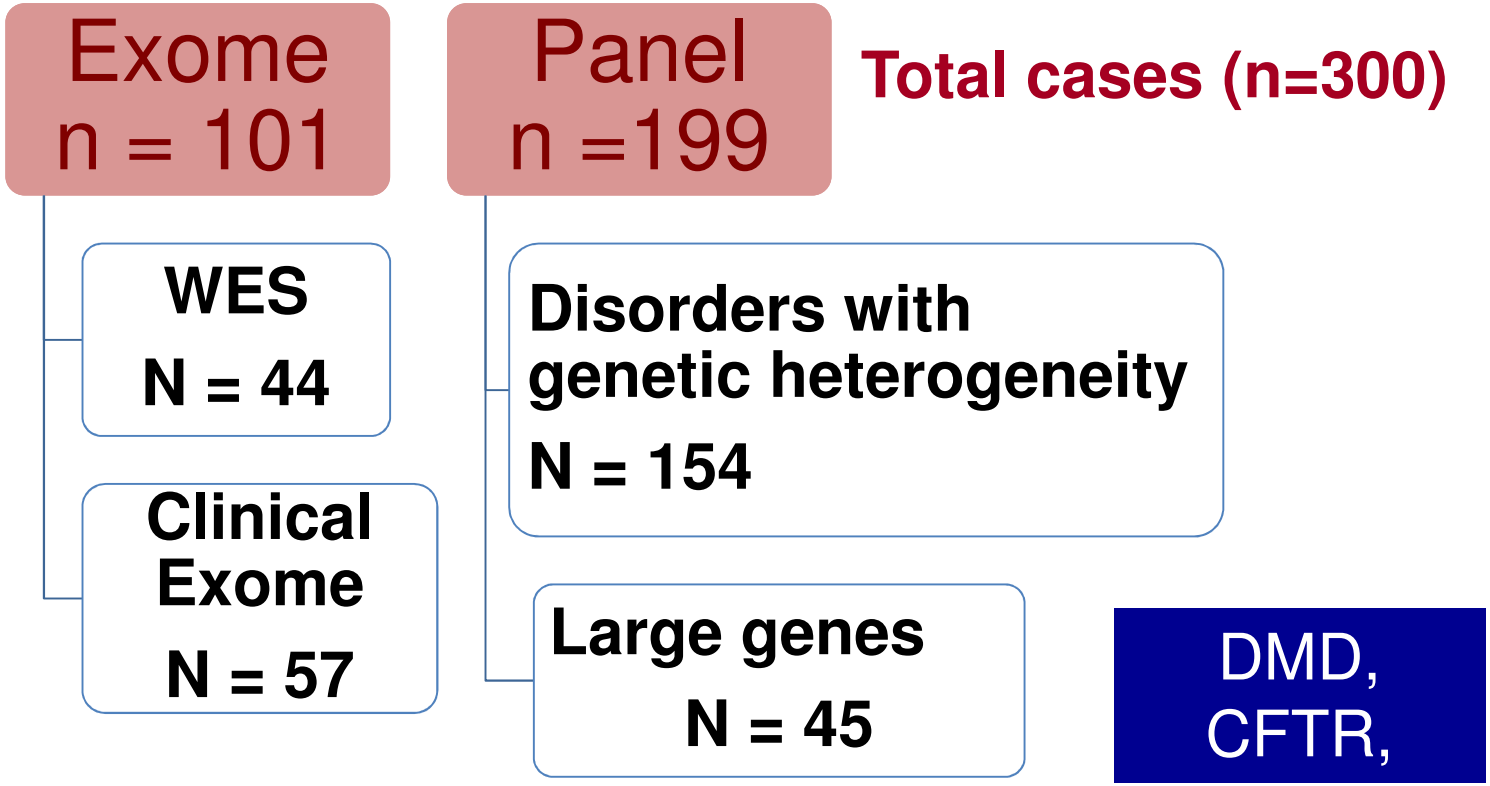
Genetics Clinic: average of 4000- 4500 patients per year

- Reproductive genetics / Neurogenetics
- Cardiogenetics / Metabolic genetics



Indications for Exome

- Neurological
- Congenital anomalies
- Neuromuscular
- Metabolic
- Misc.

Results for Exome

- Diagnosis established in 55% cases
  - In 22% clinical diagnosis was not definite at the time of testing
  - In 33% the analysis was performed in view of genetic heterogeneity

- In India, exome sequencing is being carried out by many labs and used by multiple specialists & investigators
- No data sharing between labs / investigators
- Routinely not using Phenotips / HPO
- Need to classify the phenotype better
- We find a large no of VOUS – not in the western data
- Need to characterize the variants in the Indian population that are different
- Need to collaborate to list these variants

- Share to find new genes and phenotypes
- Create a nodal center for collaboration
- Need for all stakeholders to collaborate

Thank you