Genetic mechanism of rare neurological disorders

최 무 림

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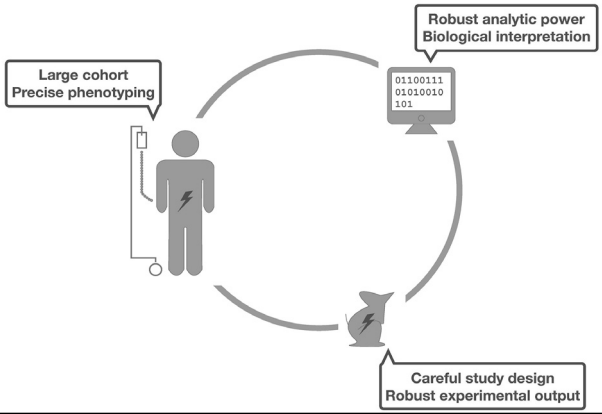
Overview

- Neurodevelopment disease cohort (*n* = 553)
- Neuromuscular disease cohort (*n* = 107)

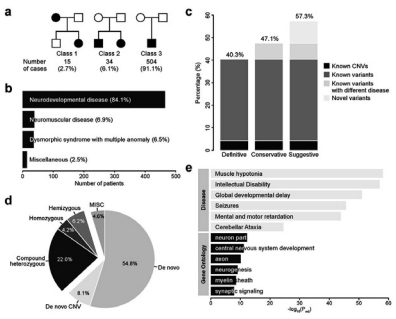
Overall approach



Overview

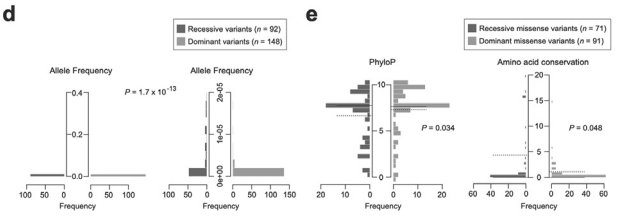
- Neurodevelopment disease cohort (*n* = 553)
- Neuromuscular disease cohort (*n* = 107)

What we've learned so far - 553 patients with ND problems



이영하 *et al., submitted*

Recessive diseases: variants vs genes?



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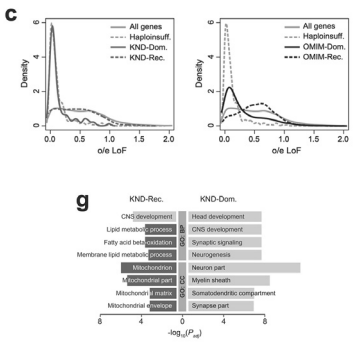

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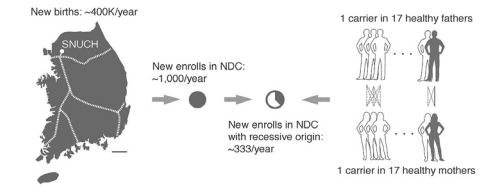
Can we predict disorders in a recessive pattern?



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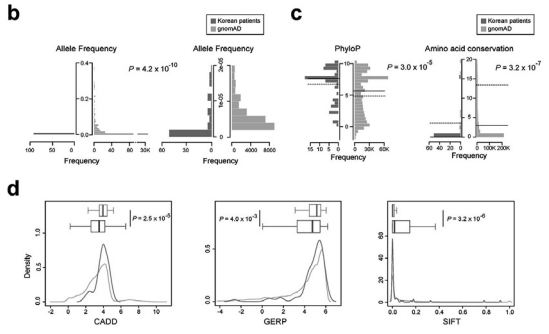

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Neuromuscular diseases

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Study Hypothesis

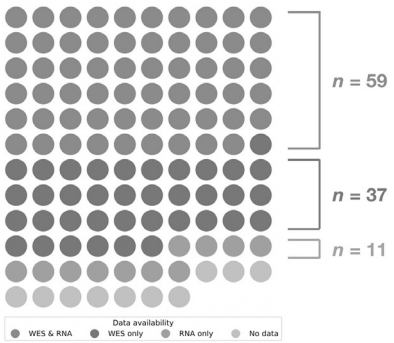
- Can we increase diagnostic rate and disease mechanism by combining genome and transcriptome data?

Pros and Cons

1. Tissue accessible
2. Support from histology
3. Recessive variants
4. Understudied

1. Rare
2. Heterogeneous
3. Structural genes
4. Understudied

Study Overview



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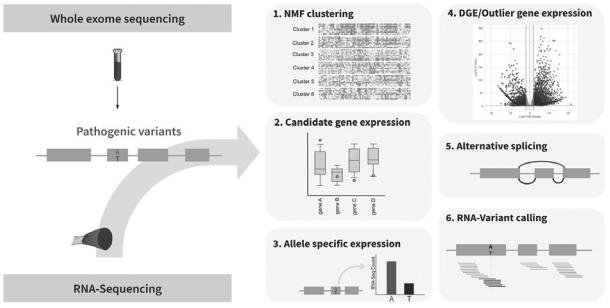
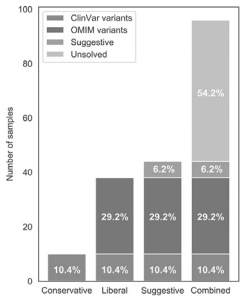


Genome analysis



- 54% of the samples still do not have readily usable variants after WES

Overall summary

1. WES based rare disease analysis shed new insights into disease mechanism and variant profile
2. RNA-seq and WGS augment WES-based analysis
3. Novel genes provide new revenue for additional benefit to the researchers and patients