

GENETIC EPIDEMIOLOGY OF ALS IN NORWAY

A 2-YEAR POPULATION BASED STUDY



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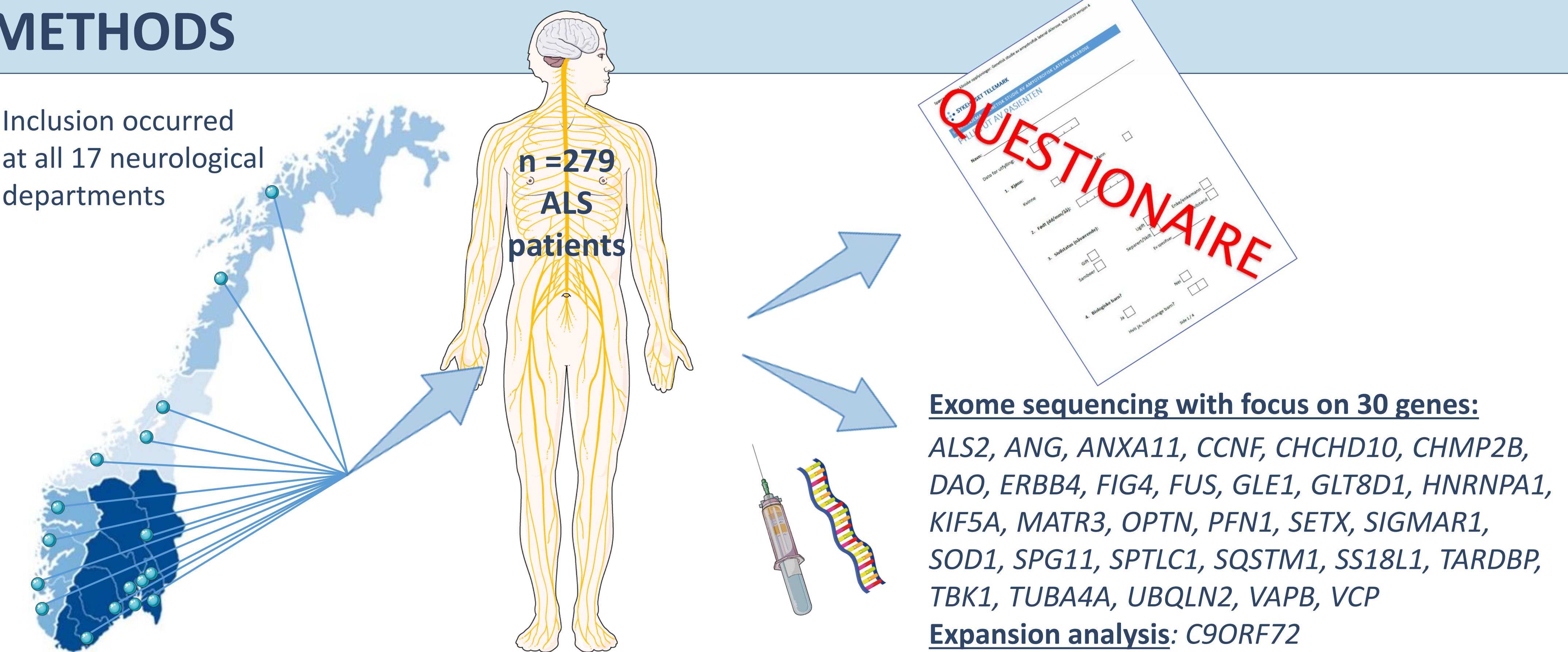
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BACKGROUND

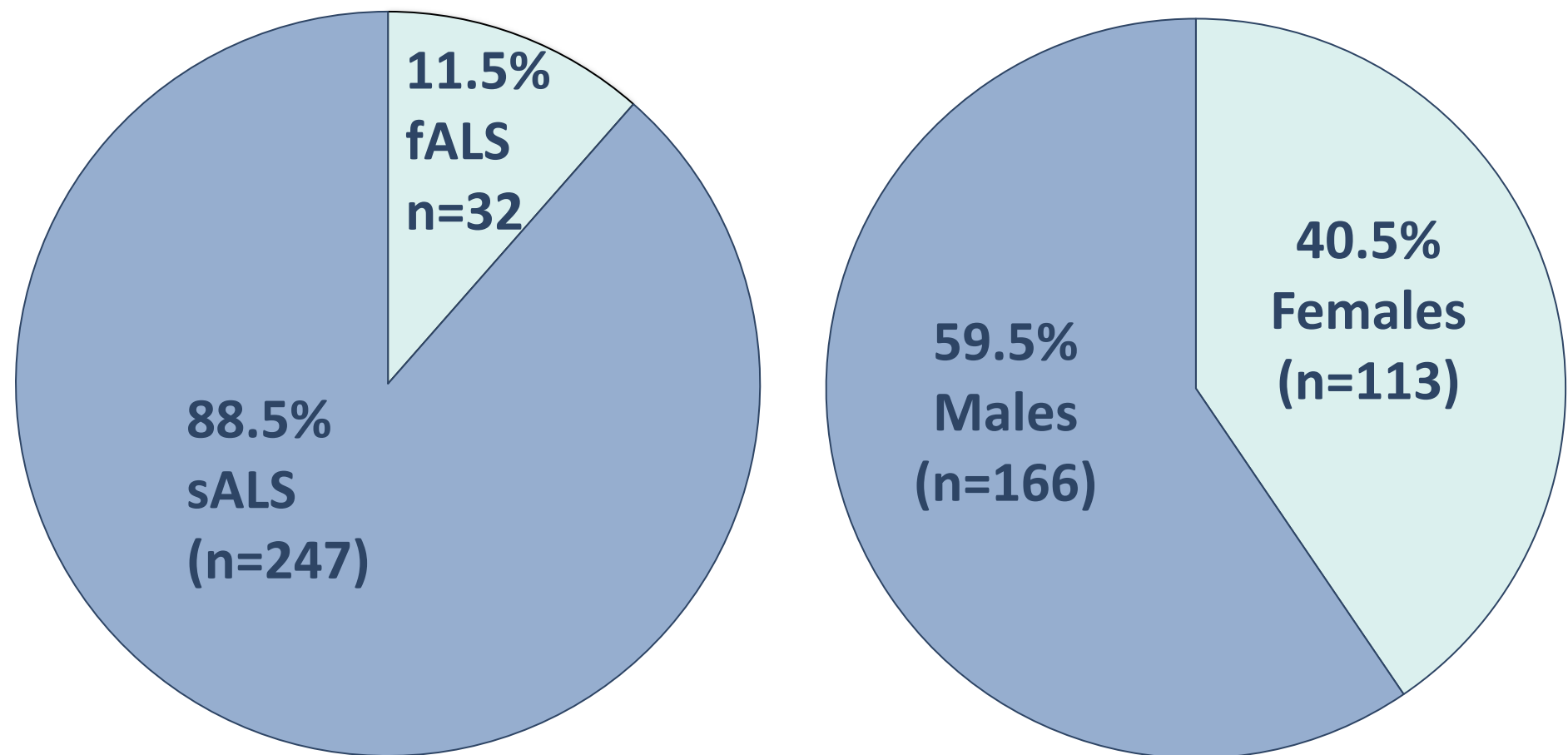
- Our research on ALS aims to:
- Provide data on the genetic factors causing ALS in Norway.
 - Increase knowledge regarding genetic diagnostics and counseling in the Norwegian health care system.
 - Give an estimate on the number of individuals that might be eligible for gene-specific clinical trials in the near future.

METHODS



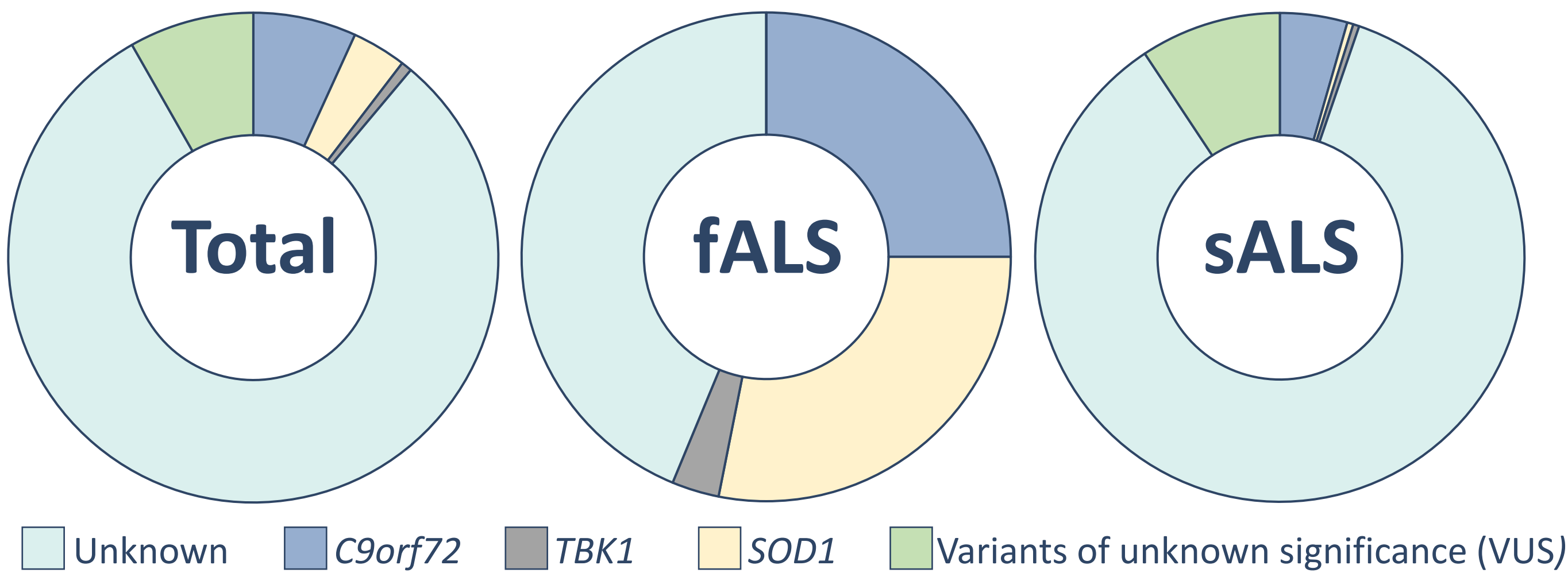
RESULTS

Clinical Characteristics



Clinical characteristics	Total	fALS	sALS
Age at onset, mean (95% CI)	62 (61-63)	58 (53-63)	62 (61-64)
Site of onset			
Bulbar	23.6%	19.4%	24.1%
Spinal	62.7%	74.2%	61.2%
Both	13.8%	6.5%	14.7%

Identified genes



Gene	Total	fALS	sALS
C9orf72	19 (6.8%)	8 (25.0%)	11 (4.5%)
SOD1	10 (3.6%)	9 (28.1%)	1 (0.4%)
TBK1	2 (0.7%)	1 (3.1%)	1 (0.4%)
VUS	23 (8.2%)	0 (0%)	23 (9.3%)
Unknown	225 (80.6%)	14 (43.8%)	211 (85.4%)

CONCLUSION

- A genetic cause of ALS was identified in 31 individuals (11.1%), 18 fALS / 13 sALS.
- The most common genetic cause was the C9orf72 expansion. Pathogenic or likely pathogenic variants of SOD1 and TBK1 were identified.
- Restricting genetic analysis to only fALS cases would miss more than 40% of those with a disease-causing genetic variant, indicating the need for genetic analysis in sALS cases as well.

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