Changing the landscape of ALS in Africa: Accomplishments

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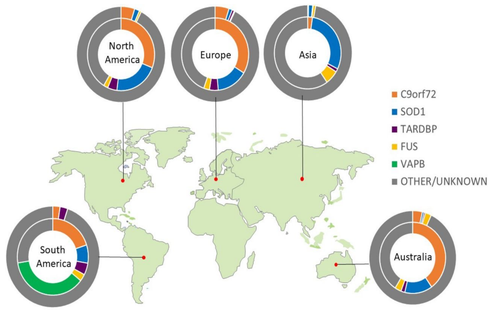
Background

- ALS has been a neglected disease in Africa
- The ALS phenotype in African patients is poorly described, with *no* data on behaviour and cognition
- There is very limited data on the genetic profile of ALS in Africa which is important for gene specific therapies

ALSA seed grants 2018 & 2022: AIMS

- Adapt and norm the Edinburgh Cognitive ALS Screen (ECAS) in Afrikaans, SA English, isiXhosa
- Organize 4 ALS research centres in Africa and establish the ALSAfrica network



Global genetic architecture of ALS. No data for Africa

Dharmadasa et al. Pract Neurol 2022

The Aga Khan University Hospital, Nairobi

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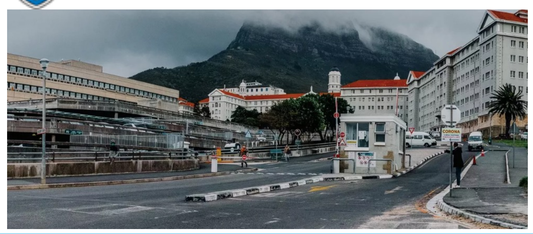
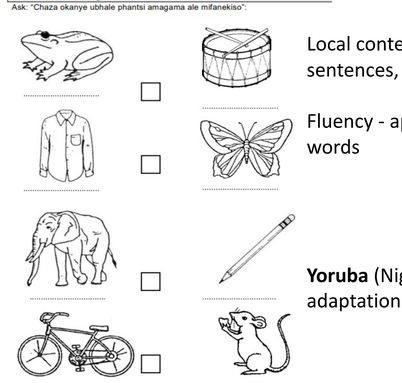


UK English

Adaptation and norming of the Edinburgh Cognitive and behavioural amyotrophic lateral sclerosis screen (ECAS) for three language groups in South Africa

Christine H. Albertyn, Anneli Hardy, Leonhard A. Bakker, Makhaya Hlangani, Kristien Van Der Walt, Braam Zeilinga, ...show all



Local content for story, sentences, spelling

Fluency - appropriate words

Yoruba (Nigeria)– adaptation "norming"

Methods

Clinical and behavioural data captured on standardized REDCap

ECAS translated to isiXhosa, Afrikaans, SA English and Yoruba

Whole blood shipped to Cape Town for DNA extraction

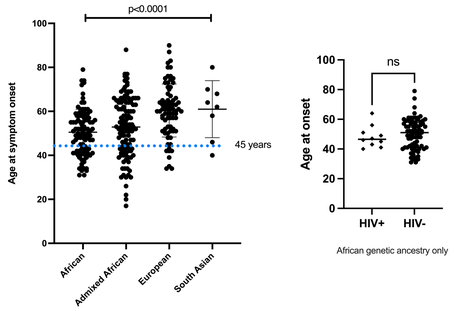
Phase Ia: Whole exome sequencing & ALS panel gene screening

Phase Ib: A commercial *C9orf72* assay screens for expansions

Phase II: research & discovery

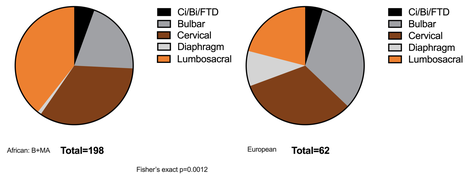
Results

Symptom onset earlier in African ancestry ALS patients



Results

Symptom onset by anatomical region varies by genetic ancestry



Frequency of *C9orf72* expansions in African ancestry ~4% compared to 7% in patients of European ancestry; while the frequency of *SOD1* mutations is 1% in Europeans vs 4% in African-genetic ancestry.[2]

Accomplishments

- Establishing research clinics changes the patient's ALS journey despite absence of therapy.
- Gathering cognitive and behavioral data in Africans with ALS
- We have changed the accessibility of Africans with ALS to obtain actionable gene results

Next steps

- Support for ALSAfrica network sustainability is required
- Much can be accomplished with seed grants

Acknowledgements

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Website

alsafrica.org

References

1 Albertyn et al. ALS Frontotemp Degen 2022
2. Nel et al. Neurology,Genet 2022