M429: Discriminating neurofibromatosis and healthy children with multimodal MRI and machine learning

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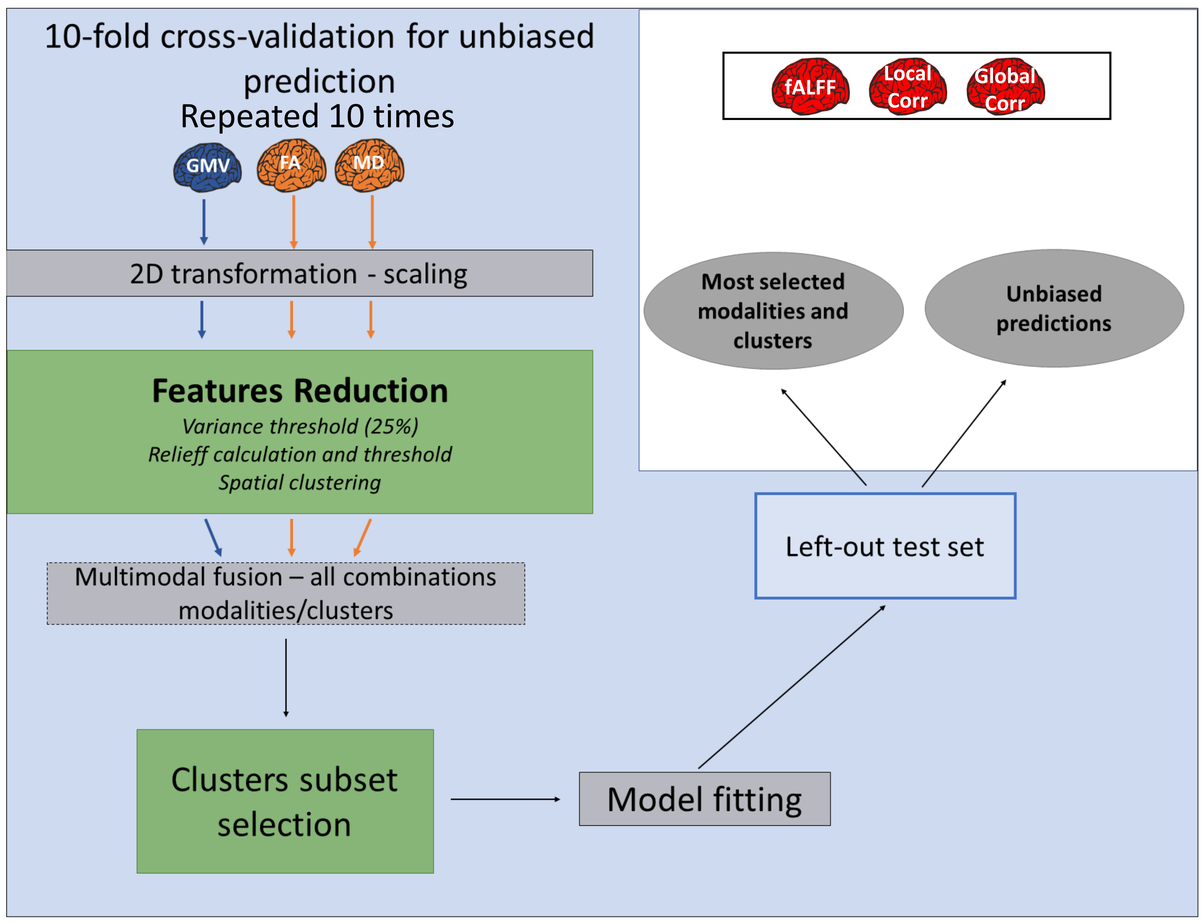
Background and Aim

Neurofibromatosis type 1 (NF1) leads to brain anomalies involving both grey and white matter. The extent and granularity of these anomalies, together with their possible impact on brain activity, is still unknown.

Although previous studies inquired grey matter volume (GMV) [1], white matter[2,3], combined grey and white matter[4], as well as functional[5,6] NF1-related abnormalities, these indexes have not been inquired together using multivariate analysis to find the multivariate signature of NF1 brain pathology.

The aim of this study is to combine indexes pertaining grey matter, white matter and brain function at rest to discriminate between NF1 and typically developing children (TD) by means of a multivariate pipeline.

Materials & Methods[7]



- 80 children (42 TD, 38 NF1): Toulouse 21 TD/21 NF1, Marseille 21 TD/17 NF1
- TD: mean age = 121.2 (± 13.9) months (20 F)
- NF1: mean age = 115.3 (± 16.5) (23 F)

: indexes[indexes from the same MRI modality share the same color]

: indexes from fMRI ; have only been tested in isolation

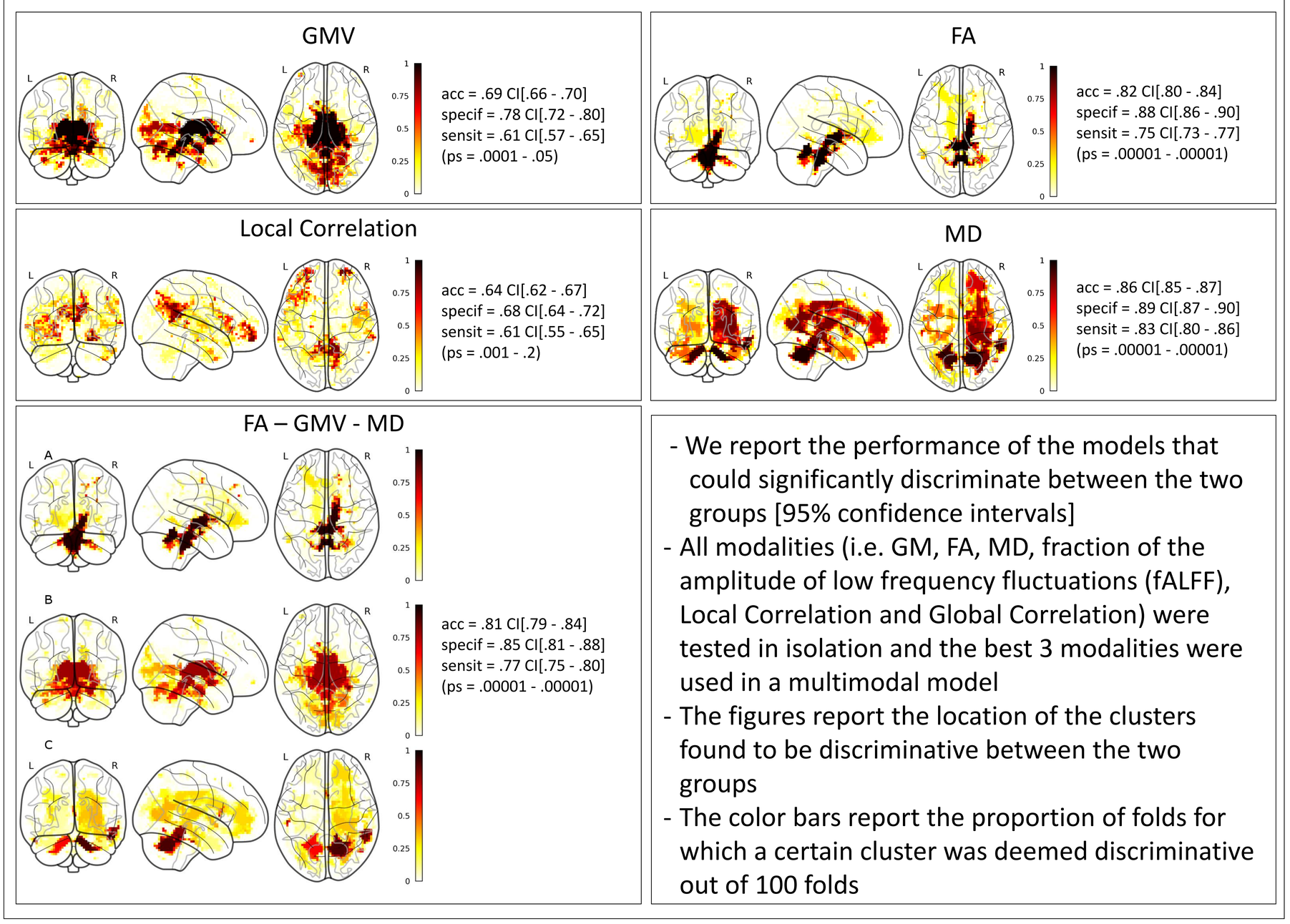
: processing steps and end-points

: features reduction/selection steps

: multimodal fusion, performed only for the model including all structural indexes (GMV, mean diffusivity (MD) and fractional anisotropy (FA) together)

All results were corrected for movement during acquisition, total intracranial volume, sex and age, and were independent from the presence of unidentified bright objects.

Results



- We report the performance of the models that could significantly discriminate between the two groups [95% confidence intervals]
- All modalities (i.e. GM, FA, MD, fraction of the amplitude of low frequency fluctuations (fALFF), Local Correlation and Global Correlation) were tested in isolation and the best 3 modalities were used in a multimodal model
- The figures report the location of the clusters found to be discriminative between the two groups
- The color bars report the proportion of folds for which a certain cluster was deemed discriminative out of 100 folds

Conclusions

- Multimodal MRI can discriminate between NF1 and TD children
- The most discriminative features are the grey matter volume, the fractional anisotropy and the mean diffusivity
- The localization of the most discriminative clusters for GMV, FA and MD are in line with previous results[1,2,3,4]
- MD abnormality seems to be particularly wide-spread and alone can discriminate as well as the three combined structural indexes between NF1 and TD children
- The three structural indexes (e.g. GMV, FA and MD) bring complementary information
- fMRI-related indexes seem to be less central to NF1 brain signature

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