“Discriminating between neurofibromatosis-1 and typically developing children by means of multimodal MRI and multivariate analyses”.

F Nemmi, F Cignetti, C Assaiante, S Maziero, F Audic, P Péran, Y Chaix EAN 2019, Oslo – Oral Presentation O2211

Background and aims: Neurofibromatosis type 1 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. Methods: In this multicentric cross-sectional study we submitted a sample of 42 typically developing and 38 neurofibromatosis-1 children to a multimodal MRI assessment including T1, diffusion weighted and resting state functional sequences. We used a pipeline involving several features selection steps coupled with multivariate statistical analysis (supporting vector machine) to discriminate between the two groups while having interpretable models. We used MRI indexes measuring macro (grey matter volume) and microstructural (fractional anisotropy, mean diffusivity) characteristics of the brain, as well as indexes of brain activity (fractional amplitude of low frequency fluctuations) and connectivity (local and global correlation) at rest. Schematical representation of the multivariate pipeline
Results: We found that structural indexes could discriminate between the two groups, with the mean diffusivity leading to performance as high as the combination of all structural indexes combined (accuracy=.79), while functional indexes had worse performances.
Conclusion: The MRI signature of NF1 brain pathology is a combination of grey and white matter abnormalities, as measured with grey matter volume, fractional anisotropy and mean diffusivity.