"SPAZIO HUNTINGTON – A PLACE FOR CHILDREN": AN ITALIAN OBSERVATIONAL, MULTICENTRE, PROGRAM TO DETECT PEDIATRIC HUNTINGTON DISEASE CASES

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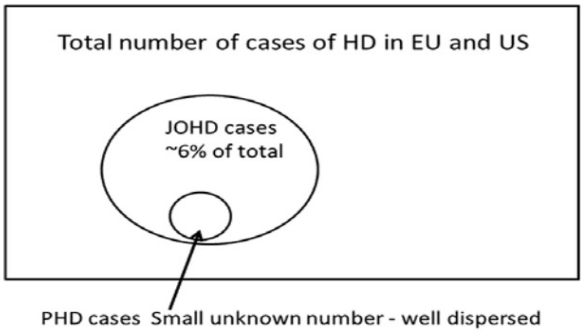
Background: The pediatric HD (PHD) variant differs from adult HD for clinical, genetic, neuropathological and imaging aspects. We have been conducting a family-based study since July 2019.
Aims: We provide a clinical description with neuropsychological and developmental profiles of 4 PHD patients.

Methods: In 2019, the "Spazio Huntington" program was launched by the Italian League for Research on Huntington (LIRH) Foundation, to increase the family awareness on children at risk of HD, potentially including PHD cases. Whenever there were "red flag" signs and symptoms of PHD, an *ad hoc* protocol was applied, including brain MRI, developmental and neuropsychological evaluation and general pediatric assessment with growth and nutritional assessment. Genetic test was performed only in children with suspicious PHD signs, after parents written consent and according to the International Guidelines for genetic testing in minors.

Results: The screening of 20 HD families with at risk minors that we met in a nonmedical environment revealed 5 children with suspicious PHD. Four children had a confirmed diagnosis with highly expanded CAG mutation and paternal inheritance. The main phenotype was a progressive gait disturbance with predominant dystonic postures, no chorea, history of developmental delay with abnormal eye movements and a coordination disorder. The age of onset ranged between 2 and 4 years. Behavioral and cognitive abnormalities included outbursts, affective problems, low impulsivity control and mild intellectual disability. EEG abnormalities with or without seizures were a common feature. Brain MRI confirmed the atypical brain pattern with a bilateral, reduced, striatum volume in absence of cortical or white matter atrophy.



Adapted from Quarrell Movement Disorders, 2019

Conclusions: We confirm the atypical PHD presentation and brain imaging pattern. The Spazio Huntington family-based approach allowed the disclosure of early PHD diagnosis and may represent a strategy to recruit children in further clinical trials.

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EHDN European Huntington's Disease Network – Advancing Research, Conducting Trials, Improving Care