The new Wilson's like disease: a treatable metabolic manganese disorder causing Parkinsonism and dystonia

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A new metabolic genetic condition related to manganese accumulation into the brain and particularly into basal ganglia has been described some years ago and was characterized mainly in early infantile and child age by dystonia and in adult by dystonia parkinsonian syndromes. This condition was due to genetic mutation in a manganes transport protein able to transfer manganese from tissue outside the cells and eliminates trough urines. Metal accumulation is evident by MRI showing metal deposits mainly in basal ganglia. We will described our experience on this syndrome, describing the clinical phenotype in childhood and adulthood, the MRI abnormalities and the effect of kelating treatment by EDTA on manganese level and on the clinical symptoms. Some experimental data will be also reported investigating the in vitro apoptotic process in cultured fibroblast showing that this process is abnormally regulated in the patients samples if related to normal subjects.