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Sommario/riassunto

Research on the multiple aspects of cognitive impairment in Down syndrome (DS), from genes to behavior to treatment, has made tremendous progress in the last decade. The study of congenital intellectual disabilities such as DS is challenging since they originate from the earliest stages of development and both the acquisition of cognitive skills and neurodegenerative pathologies are cumulative. Comorbidities such as cardiac malformations, sleep apnea, diabetes and dementia are frequent in the DS population, as well, and their increased risk provides a means of assessing early stages of these pathologies that is relevant to the general population. Notably, persons with DS will develop the histopathology of Alzheimer’s disease (formation of neuritic plaques and tangles) and are at high risk for dementia, something that cannot be predicted in the population at large. Identification of the gene encoding the amyloid precursor protein, its localization to chromosome 21 in the 90’s and realization that all persons with DS develop pathology identified this as an important piece of the amyloid cascade hypothesis in Alzheimer’s disease. Awareness of the potential role of people with DS in understanding progression and treatment as well as identification of genetic risk factors and also protective factors for AD is reawakening. For the first time since DS was recognized, major pharmaceutical companies have entered the search for ameliorative treatments, and phase II clinical trials to improve learning and memory are in progress.

Enriched environment, brain stimulation and alternative therapies are being tested while clinical assessment is improving, thus increasing the chances of success for therapeutic interventions. Researchers and clinicians are actively pursuing the possibility of prenatal treatments for many conditions, an area with a huge potential impact for developmental disorders such as DS. Our goal here is to present an overview of recent advances with an emphasis on behavioral and cognitive deficits and how these issues change through life in DS. The relevance of comorbidities to the end phenotypes described and relevance of pharmacological targets and possible treatments will be considerations throughout.Research on the multiple aspects of cognitive impairment in Down syndrome (DS), from genes to behavior to treatment, has made tremendous progress in the last decade. The study of congenital intellectual disabilities such as DS is challenging since they originate from the earliest stages of development and both the acquisition of cognitive skills and neurodegenerative pathologies are cumulative. Comorbidities such as cardiac malformations, sleep apnea, diabetes and dementia are frequent in the DS population, as well, and their increased risk provides a means of assessing early stages of these pathologies that is relevant to the general population. Notably, persons with DS will develop the histopathology of Alzheimer’ s disease (formation of neuritic plaques and tangles) and are at high risk for dementia, something that cannot be predicted in the population at large. Identification of the gene encoding the amyloid precursor protein, its localization to chromosome 21 in the 90’s and realization that all persons with DS develop pathology identified this as an important piece of the amyloid cascade hypothesis in Alzheimer’s disease. Awareness of the potential role of people with DS in understanding progression and treatment as well as identification of genetic risk factors and also protective factors for AD is reawakening. For the first time since DS was recognized, major pharmaceutical companies have entered the search for ameliorative treatments, and phase II clinical trials to improve learning and memory are in progress. Enriched environment, brain stimulation and alternative therapies are being tested while clinical assessment is improving, thus increasing the chances of success for therapeutic interventions. Researchers and clinicians are actively pursuing the possibility of prenatal treatments for many conditions, an area with a huge potential impact for developmental disorders such as DS. Our goal here is to present an overview of recent advances with an emphasis on behavioral and cognitive deficits and how these issues change through life in DS. The relevance of comorbidities to the end phenotypes described and relevance of pharmacological targets and possible treatments will be considerations throughout.