Down syndrome is a genetic syndrome related to trisomy 21, and characterized by intellectual and adaptive deficiencies, facial deformities, cardiopathies, and hypotonia that determine a specific cognitive-behavioral phenotype. The behavioral and psychiatric cognitive phenotype and its evolutionary profile impose bioethical considerations in the down to promote better and personalized clinical and relief, diagnostic and therapeutic strategies to favor an adequate insertion of the down in the scholastic and work environment.

**Keywords:** cognitive disabilities, bioethics, Down syndrome.

**DOI:** 10.19193/0393-6384_2019_6_551

**Received** November 30, 2018; **Accepted** February 20, 2019

**Introduction**

The Diagnostic and Statistical Manual of Mental Disorders 5 (DSM-5), published by the APA (American Psychiatric Association) in 2013, is a manual of classification of mental illnesses. DSM-5 defines intellectual disabilities as “neurodevelopmental disorders” that begin in childhood.

**DSM-5 diagnosis of intellectual disabilities follows these criteria:**

- Deficits in intellectual functioning - reasoning, abstract thinking, planning and learning - confirmed by clinical evaluation and individualized standard testing.
- Deficits in adaptive functioning - conceptual, social, and practical skills.

Adaptive functioning relate to the ability to carry out the most important age-appropriate daily needs - in the social and cultural environment - concerning self-care standard.

Deficits in adaptive functioning lead to limited communication and social skills and participation in family, school and job activities.

**Intellectual disabilities in Down syndrome**

Intellectual disabilities are diagnosed by the 4-5 years of age through standard methods that measure the global impairment - intellectual and adaptive - of the child. In some genetic syndromes - like Down syndrome - intellectual and adaptive deficits occur that can lead to a specific cognitive-behavioral phenotype. Down syndrome (DS), described in 1866 by Langdon Down in London and by Edward Seguin in New York, has been linked to trisomy 21 since 1959.

DS is associated to intellectual and somatic deficits, characteristic facial features, congenital heart disease, gastrointestinal and genitourinary disorders, hypothyroidism, cataract, glaucoma, and, since birth, hypotonia and eating disorder.

The evolution of the psychomotor development is slowed down in both the motor performances and the cognitive and language development$^{[1,2,3]}$.

The cognitive profile is characterized by mild or severe deficits$^{[4,5,6]}$, with the mean IQ of 50 (ranging from 30 to 70) and the adaptive level is consistent with mental age$^{[7]}$. 
The cognitive profile features better visuospatial skills than verbal skills, even if, later in life, a cognitive impairment occurs\(^8\).


Language development shows, at first, verbal production and comprehension comparable to other children with the same mental age\(^9\). However, in the following years, a language deficit can be noted, with a gap between vocabulary and morpho-syntax skills\(^4,10\).

Hearing deficits, often found in these patients, may worsen even more already present speech disorders\(^11\). During school age, verbal fluency highlights a simple syntax, but the comprehension is adequate for the development profile\(^12\).

Difficulties in verbal fluency are phono-articulatory\(^13\) and to compensate the language deficits, gestures develop before language, using hand signs efficiently\(^14\).

In children with DS, the verbal fluency deficit can be compensated by a communication therapy, the sign language, to strengthen their social and communication skills.

Subjects with DS, during their youth, may present psychiatric disorders\(^15,16,17,18\).

Such disorders could be attention disorders and impulsive behavior\(^19,20\); anxiety, obsessive-compulsive disorders and depression, that is often associated with somatic disorders (e.g., changes in appetite, sleep disorders, concentration difficulties)\(^21\).

Often, stressful events happen before the onset of depressive symptomatology\(^22\).

Obsessive-compulsive disorders may exhibit repetitive behavior, like polydipsia and hyperventilation. Psychiatric disorders are treated in these subjects with cognitive behavioral psychotherapy, related to the subject’s cognitive skills\(^23\).

In the subjects with DS, the drug therapy includes selective serotonin reuptake inhibitors (SSRI), with an antipsychotic as adjuvant in the event of hallucinations.

In these subjects, cognitive skills are prone to get better with time, slowly, up to adulthood, during which dementia may occur\(^24\).

The subjects with DS may develop progressive cognitive impairment resembling, over time, an Alzheimer-type dementia (AD).

DS and AD share neuropathological and neurochemical defects linked to chromosome 21\(^25\).

Many genes are linked to neurodegenerative mechanisms, among which the Cu-Zn superoxide dismutase 1 (SOD1), the Ets2 transcription factor, and the APP (Amyloid Precursor Protein), all located on chromosome 21\(^26,27\).

An elevated expressivity of the APP gene would favour the accumulation of B-amyloid plaques, a neurodegenerative phenomenon characteristic of AD. AD may occur in 25% of the subjects with DS after age the of 40 and the percentage increases with age up to 60% between the age of 50 and 60\(^28,29,30,31\).

**Bioethical implications**

The cognitive-behavioral and psychiatric phenotype and its evolution profile require some bioethical considerations concerning DS.

For starters, it is important to emphasize the new scientific contributions, to study better the cognitive-behavioral impairments and a better therapeutic role of the more appropriate and efficient medication to use.

A more detailed study on the cognitive, behavioral and socio-relational profile of the subjects with cognitive disabilities in genetic syndromes like DS may allow to improve diagnostic processes and to promote better intervention strategies, more and more customized, as regards the fundamental bioethical principles of “beneficence” and “non-maleficence” that follow and improve every clinic, diagnostic and therapeutic strategy in the rehabilitation, scholastic, social and work fields.

It is crucial, thus, that the bioethical considerations and conclusions, in order to treat and guide a child, from the start, and after throughout all their evolutionary growth process, should always target the neurobehavioral weaknesses of the children suffering from DS. Thereby, in order to achieve ethical choices useful for the clinic profile, that guide the diagnosis, the therapies, the rehabilitation and the treatment of neurobehavioral disorders, during each stage of development in the school age and adulthood.

**Conclusions**

The bioethical conclusions concerning genetic syndromes with neurobehavioral profiles characterised by intellectual and adaptive deficits must show a better diffusion of scientific field knowledge, for the doctors and the healthcare workers, and better communication from them, adequate for the families and to make them aware of the onset and evolution of
intellectual and adaptive impairments. Everyone has to be a part of the implementation of the best therapeutic and rehabilitative strategies in the subjects with DS, to integrate better these subjects in their familiar, scholastic, social and work environment.

References

7) Chapman R, Hesketh LJ. Behavioural phenotype of individual with down syndrome, mental retardation and developmental disabilities research reviews 2002; Vol. 6, 84-95.