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DOWN SYNDROME AND AUTISTIC DISORDER: AN “ITALIAN CASEWORK”

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ABSTRACT

Autism is a complex brain disorder resulting in social, behavioural and language problems. It is characterized by impairments in language and nonverbal communication, deficits in social interaction, and restricted, repetitive, stereotyped patterns of behaviour, interest, and activities. Other factors can be associated with or found in conjunction with autism. These include conditions such as variable cognitive impairments, attention deficit hyperactivity disorder, anxiety, obsessive-compulsive disorders, and Down syndrome. The reason for the development of autism in Down syndrome people is not apparent. The behavioural features of autism may result from differences in brain structure or function, which directly affect the presence of Trisomy 21. It is also possible that the increased rate of autism will be secondary to the increased vulnerability of infants with Down syndrome. In this work, a casework is discussed.

KEYWORDS: Down Syndrome, Autism, Rehabilitation Treatment, Educational Programming.

INTRODUCTION

The diagnosis of autism is quite tricky and becomes even more uncertain in people with Down syndrome or other known forms of intellectual disability. Several studies (Ghaziuddin, Tsai & Ghaziuddin, 1992; Ghaziuddin, 1997; Wing & Gould, 1979; Wakabayashi, 1979; Howlin, Wing & Gould, 1995; Rondal & Guazzo, 2021) have found that although there is no epidemiological study of the prevalence of autism-type disorders in a large population of Down's syndrome children, what is usually apparent is that the incidence is in the 10% range. It is difficult to have accurate data; many cases are undiagnosed or are diagnosed at an advanced age. In the case of Down syndrome, the parents of a child c the same caregivers are busy with the various clinical-medical problems (heart disease, hypotonia, altered thyroid function, joint laxity, visual impairment, etc.) typical of the syndrome and with the multiple aspects of the expected developmental delay (deficits in gross-fine-motor, communicative, sensory, performance skills, etc.) and, precisely because of this expected delay, the possibility that one may also be in the presence of autism is not taken into consideration. Moreover, many of the typical symptoms of autism may be considered characteristic of Down syndrome and not sufficient to diagnose autism (Maltz, Schopler, 1979; Guazzo & Acampora, 2006; Pary, 1997).

The area that is most impaired during the development of the Down child is, in fact, the cognitive area, i.e., the development of cognition (thinking, memory, reasoning, etc.) and language (expression and comprehension), and it is precisely in these areas of development that a delay is expected. Another area of development is the social-emotional area: this area has more normal development in a Down syndrome child than in an autistic child (Loveland & Kelley, 1988, 1991).

Most Down children show a slight delay in social-emotional development; smiling as a response comes at about 2 months (out of an average of 1.5-4 months), spontaneous smiling at 3 months (out of an average of 2-6 months), and parental recognition at 3.5 months (out of an average of 3-6 months), each of these milestones is reached only one month behind the average. Although some studies state that the intensity of affective responses such as smiling and laughing may be slightly less than that exhibited by a typically developing child, parents respond warmly to the onset of smiling and eye contact. Down children begin to take pleasure in small games such as "What hand is it in" or "cu-cu-sette" at about 11 months (average 9 to 16 months), which means a 3-month delay compared with typically developing children. Studies done during the second year of life show that children have social communication skills; they even know how to use social skills to distract an adult from a task they do not want to tackle. Down children are endearingly affectionate and respond generally to physical contact, unlike autism.

Thus, when language appears and ambulation begins, new communicative perspectives open up, and more complex forms of social behaviour can be implemented. Play behaviour and the evolution of "detachment" (Gesell, 1965) have greater significance at this stage.

A particularly significant signal for early social interactions relates to gaze. Typically, it is directed very early toward the human figure in developing children. In particular, the infant's interest is captured by the face from the first month of life. Gunn, Berry, and Andrews (1982) found that Down children, during face-to-face interactions with their mother, spend most of their time looking at their mother (interpersonal gaze) rather than shifting their gaze to their surroundings (reference gaze). Berger and Cunningham (1986) observed that Down children need more time to develop sustained attention toward adults. Motti, Cicchetti, and Sroufe (1983) argued that older Down children develop visual attention toward objects but do not readily engage adults in play with objects. The results of research by Suson and Wetherick (1981) on a sample of three- to four-year-old children with Down syndrome confirm that Down children cannot precisely maintain eye contact and mutual gaze, which is the cause of their isolation within a playgroup. The absence of eye contact is interpreted by other children as an unwillingness to interact, despite the authors finding average social skills for the Down group in the research.

This normal emotional response persists into adult life and, as studies of adolescents have shown, develops into proper empathy, which makes Down people, people who are sensitive and attentive to the needs of others, people with whom it is pleasant to live.

The key points, therefore, to be examined in a child with Down syndrome in whom behaviour disorders, typical of autistic disorder, are suspected are (Guazzo & Acampora, 2006):

1. High isolation level: the child does not relate to others typically and seems to prefer being left alone. The child seems to regard people as objects, not human beings. He does not join playgroups with other children, and unlike Down children, who are generally very affectionate and expansive, the autistic child does not want to be hugged.
2. Anxious and obsessive desire to preserve his or her status: any change in the daily routine can cause a big mood.
3. Lack of eye contact: it is typical for autistic people not to have eye contact; they do not look at people or beyond the other person.
4. Presence of repetitive and "stereotyped" motor behaviours: e.g., sitting with an object in their hand, moving it back and forth and staring at it.

Children with Down syndrome to whom the diagnosis of autism can be added generally show deficits in these areas, as can also be inferred from our case described in the next section.

Rehabilitation treatment of a Down syndrome person with autistic disorder: the case of Luca

Six-year-old Luca began Physiotherapy rehabilitation treatment at the age of two with the diagnosis of: 'Psychomotor Developmental Delay in Subject with Down Syndrome' later, at the age of four, he began a rehabilitative treatment of Psychomotricity and Speech Therapy, and the diagnosis was changed to 'Psychomotor Delay with Affective Immaturity and Social Isolation in Subject with Down Syndrome'.

Luke, at the first observation about three years ago, presented a whole series of maladaptive behaviours: he was constantly fidgeting, his requests had to be fulfilled immediately, he was always on the move, he was excitable and impulsive, he maintained eye contact with the task for about 5s/20s (v.n.: 15s/20s); he did not establish eye contact spontaneously with interlocutors; he was not interested in the activities that were proposed to him, he used some behavioural stereotypes. The relationship with the object and its respective functionality was absent: in a few minutes, he grabbed everything within his reach, putting it away again immediately afterwards. The child did not accept any body contact and had gestural and behavioural stereotypies (he waved his hands and arms and used small toys by twirling them between his fingers). He did not explore space. In addition, he preferred to be left alone and did not accept changes in his daily routine. Imitation was almost completely absent, and he had low frustration tolerance. Clinically, these behaviours could be attributed:

- To the presence of deficits in the mechanisms that lead to the regulation and consistent maintenance of behaviour over time;
- to difficulty in inhibiting, initiating and maintaining a behaviour or response according to the demands of the task or situation;
- to accentuate difficulties when the consequences of the behaviour were deferred, weak or nonexistent.

In addition, the child presented generalized anxiety that predisposed him negatively in dealing with new or pseudo-new situations (perceived as such), affective inhibition, psychic closure, and relational disturbance.

Spontaneous speech was absent; he emitted a single word, 'pahaa' (daddy) which he uttered whenever he experienced an uncomfortable condition; however, in some cases, it was compensated by gestures, while nonverbal communication was restricted to motor behaviours, such as pulling the interlocutor, but ultimately lacked the indicative function toward objects or people; comprehension was reduced to very few sentences and, many times, was aided by gestural prompts (Rondal & Guazzo, 2012, 2021).

All these observations favoured the formulation of an additional diagnosis of "Autism Spectrum Disorder".

METHOD

Assessment

For the problems presented by Luke, in addition to direct observation, the IFP (Questionnaire for the Individualization of Functioning Profile) is a criterial tool through which it is possible to identify the skills and sub-skills mastered by the person, identify the activities that the person can perform independently and satisfactorily, and formulate the goals that should be pursued and the activities that should be proposed in an enabling and educational planning (Guazzo, 2022).

Treatment

The rehabilitative treatment was initially aimed at acquiring new skills, reducing problem behaviours and acquiring prerequisites for learning and for expressive and receptive language through some intervention procedures: eye contact on demand, joint attention, joint action, pairing and incidental learning.

Programming

Problems: Watching and listening skills not yet fully mastered; coarse- and fine-motor skills not appropriate for chronological age; imitation not yet acquired; inadequacy of communication skills.

Objectives: 1. To improve watching and listening skills; 2. To make imitative patterns acquire; 3. To make fine motor patterns functional for personal autonomy; 4. To foster verbal imitation and communicative interaction.

Procedure: The following elements were considered in the planned intervention:

1. Environment. The intervention took place in the home, at the Rehabilitation Center and at the school (with supplementary modalities to the curricular ones), in a structured environment, but without excessive acoustic, olfactory and/or tactile stimuli.
2. Materials. Two types of materials were used, one prosthetic and the other stimulation. The following are considered prosthetic or facilitating materials: balloons, bars, ladders, support boards, etc. Stimulation can be given with objects appropriately varied about size, shape and the possibility of associating an acoustic component.
3. Stimuli. In addition to using the material, the stimulus should be the operator himself, a familiar and/or unfamiliar adult (the operator's voice or other acoustic stimuli).
4. Suggestions. Help, in case of difficulty, Luke having1 care to gradually attenuate such help at the appearance of active traces of the required performance (fading).
5. Reinforcements. Social reinforcers (strokes, "good Luke," etc.), sounds, music, toys, etc., and natural reinforcers (what the child wants at that moment) should be provided.
6. Duration and frequency of sessions. To achieve the objectives, the intervention time was organized into 15–20-minute sessions interspersed with playful activities that the child liked (for a maximum of 5 minutes). (N.B. A positive response should be awaited before moving on to the play activity).
7. Criterion for moving to the next objective. A 90% criterion was adopted (e.g., out of 20 successive trials, Luke had to give about 90% correct answers; if the correct answers were between 75% and 90%, one still insisted until the 90% criterion was exceeded; correct answers of less than 75% indicated that the strategy was not suitable and should be changed.

Verification

To verify the results achieved, IPF was used again by comparing the initial observation data with the verification data carried out twelve (December 2022) and twenty-four months (December 2023) after the start of treatment (Figure 1).

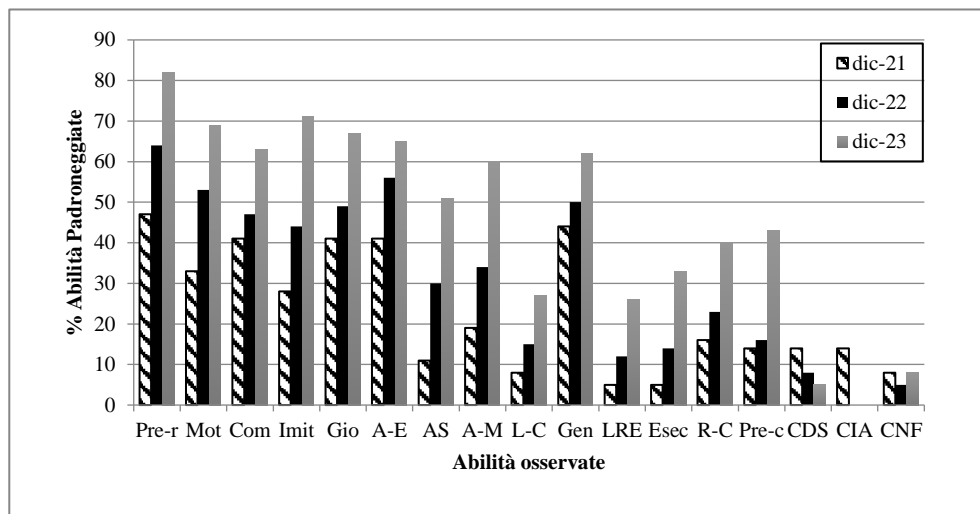


Fig. 1 - Legend: Pre-r = Pre-requisite Skills; Mot = Motor Skills; Com = Communication Skills; Imit = Imitation Skills; Gio = Play Skills; A-E = Affective-Emotional Skills, AS = Self-Sufficiency Skills; AM = Attentional and Mnestic Skills; L-C = Logical-Conceptual Skills, Gen = Generalization Skills; LRE = Receptive and Expressive Language Skills, Exec = Executive Skills, R-C = Rhythm and Coordination Skills; Pre-c = Pre-curricular Skills, CDS = Challenge Behaviors, CIA = Learning Interference Behaviors; CNF = Non-Functional Behaviors.

Intervention

During the early stages of the intervention, the child cried, struggled, refused therapy, did not dock his gaze, refused any toy and/or game that was proposed to him, could not sit up, nor could he maintain eye contact for more than 5s/20s with any task, and the comprehension of verbal language was very condition by the reinforcing presence of gestures.

The caregiver then began imitating him in ge sti and humming, attracting his attention: he would approach in curiosity and, at times, participate in games with objects and bodily involvement. As this interaction progressed, the child increasingly sought out the known objects while also becoming curious about others who were in the setting to the extent that he "dragged" the caregiver toward the object (constructions, plastic animals, ball, etc.) or desired activities (finger painting, salt pasta, shaving foam, etc.).

According to the ABA-oriented treatment, these results and the positive responses to the proposed activities convinced the operators to begin a more structured intervention to acquire the deficit prerequisite goals (Guazzo & Acampora, 2006).

Results

After the described treatment, the child had a marked reduction in stereotypies and improved gross and fine motor skills, sitting for about fifteen minutes to perform pairing, recognition, construction, manipulation, and colouring tasks.

Play experiences have also increased: he plays with the ball, kicking it or throwing it toward the operator, plays with toy cars by sliding them with wheels on the table or the floor, flies a toy aeroplane in the room, straddles a stick to "ride a horse." He also recognizes basic topological concepts: above/below, inside/outside, etc., and can "quantify" the time between a proposed task and a play activity. He indicates and sometimes verbalizes what he wants. The development of verbal imitation has also improved, and it is thanks to this that Luke can verbally express a fair number of words, about 60, which is increasing daily.

These results, together with those that can be deduced from Fig. 1, demonstrate the effectiveness of treatment through strategies geared toward the acquisition of basic skills (eye contact on demand, joint action and attention, pairing), generalized, reciprocal and delayed imitation, and language development as it is acquired by typically developing children (vocalizations, babbling, etc.), in which interaction with parents (natural reinforcers to the words 'marna' and 'pahaa' and verbal requests), interaction with peers (preschool placement) and interaction with non-parental adults (therapists and teachers) played a very important role.

So, the focus of the intervention was not directed toward the child's inadequate behaviours but on the relationship between him and the various facets of the environment in which he lived and acted. It followed that change was primarily required of the environment, which adapted to Luke's needs, changing according to the rehabilitative goals to be pursued. To the environment and the situation, where some of the causes capable of triggering many behavioural problems are often found, the "systemic" model of person-environment relationships was integrated: the 'problem' is contextual, not personal.

The system is a set of interacting units related to each other. The state of the other units constrains the state of each unit. Moreover, at least one operation can be applied to the sum of those units, which gives a value greater than the value, and that is obtained by applying that operation to the sum of those units taken individually. This view introduced within the rehabilitation universe makes it possible to highlight the close relationships of mutual influence that link the environment to the person and how an educational rehabilitation treatment can be organized.

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