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Learning Difficulties in Intellectual Disabilities: Differences Between Some Genetic Syndromes

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ABSTRACT

This article summarises the main learning difficulties in individuals with Down Syndrome (DS), Williams Syndrome (WS), Fragile X Syndrome (FXS) and Autism Spectrum Disorder (ASD). A systematic comparison of learning and functional difficulties in these four syndromes reveals the existence of partial syndromic specificity, the main theoretical and clinical implications of which are discussed.

The second part of the paper illustrates the main stages of educational and rehabilitative interventions in intellectual disabilities. An intervention is proposed into three phases: 1) early identification and frequent monitoring of students with learning difficulties, 2) pursuit of general long-term learning objectives and assessment of intermediate performance, and 3) developing coordinated and differentiated teaching interventions for all students.

Keywords

Learning Difficulties (LD), Intellectual Disabilities (ID), Down Syndrome (DS), Williams Syndrome (WS), Fragile X Syndrome (FXS), Autism Spectrum Disorder (ASD).

Introduction

The relationship between cognitive functioning and learning ability is very complex and is not limited to a simple dependency relationship, but instead refers to the interaction between highly differentiated functions: the degree and type of cognitive organisation of a child does not correlate directly with his or her academic performance [1]. For many years, the debate on Intellectual Disability (ID) has been marked by the alternative between the hypothesis of intellectual disability and that of disorder understood as cognitive atypia. It can now be argued that disability, with its dynamics, is the essential basis of cognitive atypia [2,3]: the different evolution of disability always tends to produce, over time, clinical pictures characterised by atypical intellectual organisations, related both to the depth of the deficit and to the specificity of the skills involved. Many children with intellectual disabilities have particular difficulties in passing through the various stages of development and in integrating and comparing different cognitive strategies. Some of these children, for example, tend to underuse their skills and perform below their potential. It is in this context that the relationship between intellectual disability and learning disability emerges as a central issue in the pathology of intellectual disability [1].

It is commonly believed that the key dimension in intellectual disabilities is intellectual level (IQ), possibly supplemented by an estimate of adaptive potential. Although not entirely irrelevant, this view is too general. The scientific approach to intellectual disability needs to be more aetiology-oriented. Recent advances in genetics are leading to greater attention to the behavioural phenotypes of syndromes with ID [4]. Syndromes with ID placed at similar levels on standard IQ scales have been shown to differ substantially in psychological aspects. Genetic defects (both Mendelian disorders and cytogenetic abnormalities) are involved in a substantial proportion of both milder and more severe cases of ID, suggesting that the previous equating of moderate and severe ID with a pathology, and of mild ID with normal variation (e.g., Zigler's opposition between socio-cultural and organic ID; [5,6]) is an over simplification [7]. Intervention programmes must take a greater account of the aetiological dimension and syndromic specificities.

Aetiology is becoming a significant predictive variable, and this is particularly clear for genetic syndromes leading to ID, as the initial defects in these syndromes can be identified at the chromosomal or genetic level.

As mentioned above, a cognitive functioning problem always involves a learning difficulty, the severity of which varies according to the functioning and type of intellectual disability and the specificity of which is relative to both the timing and the mode of learning. It can therefore be said that there are many intellectual and learning difficulties, variability and heterogeneity in cognitive and neuropsychological profiles, and diversity in the timing of acquisition and performance in the various manifestations (traumatic disorders, syndromes, etc.) of disability [8]. The variability of clinical pictures, in addition to highlighting some predictive parameters concerning the school difficulties of children with intellectual disabilities and identifying some characteristic stages or 'critical' moments in the learning process (such as Down syndrome, Williams syndrome, Fragile X syndrome and autism), is taken here as a starting point for better analysing the relationship between intellectual difficulties and disorders. Down syndrome, Williams syndrome, Fragile X syndrome and autism are taken here as a starting point for a better analysis of the relationship between intellectual difficulties and learning disorders.

Intellectual Disability, Learning Difficulties and Genetic Syndromes

Intellectual disability, which accounts for the most significant percentage of individuals with atypical development, is a condition that manifests itself in childhood, in which general cognitive functioning is significantly below average (IQ < 70) and is associated with a deficit in adaptive behaviour [9]. It is one of the most significant problems in compulsory education due to its frequency among learning disorders, its varying severity and expressiveness, and the difficulty of differential diagnosis with other clinical conditions, which, although starting from different aetiologies, share the same symptoms.

In this context, rather than the traditional distinction into clinical subgroups of disability, using the Intelligence Quotient (*mild*: IQ 50-70, approximately 80% of cases; *moderate*: IQ 35-49, approximately 12% of cases; *severe*: IQ 20-34, approximately 7% of cases; *profound*: IQ ≤ 20, approximately 1% of cases), a model has been preferred that responds to the need to study in greater depth the interaction between intellectual disability, development and learning: limitation to progressive change in the interaction between individuals' behaviour and events in their environment [10]. However, it is believed that all individuals with moderate, severe or very severe intellectual disabilities have an organic cause; this means that in every case, it should be possible to identify atypical brain development due to genetic or non-genetic causes that occurred during the prenatal, perinatal or postnatal period. Among the genetic causes, Down syndrome, Williams syndrome and Fragile X syndrome (Table 1) are the most widely studied. Autism spectrum disorder (ASD) can be considered a syndrome because it is a group of disorders characterised by deficits in specific areas of development, such as social interaction, communication and the presence of repetitive or restricted behaviours. Still, it is not caused exclusively by hereditary factors. Research has shown that it is a complex disorder with a strong genetic component and involving environmental factors (Table 1).

Table 1: Comparison between the four learning difficulty profiles: Down syndrome, Williams syndrome, Fragile X syndrome and ASD. The '+' sign indicates relative strength, and the '-' sign indicates relative weakness. About ASD, reference was made only to subjects at Levels 2 and 3 of the DSM-5.

LEARNING DISABILITIES	SINDROMI			
	Down	Williams	X-Fragile	ASD
Language				
- *Expressive*	-	+	-	-
- *Receptive*	-	+	-	-
- *Pragmatic*	+	-	-	-
Reading	-	+	-	-
Writing	-	+	-	-
Calculation	-	+	-	-
Metacognition	-	+	-	-

Down Syndrome

Down syndrome, also known by its aetiological name 'trisomy 21', is one of the most common genetic syndromes. The absolute frequency of the syndrome is currently around 1 in 1,000 live births. Cases of trisomy 21 are usually classified into three aetiological subcategories: 1) standard trisomy (97% of cases), 2) mosaicism (1% of cases) and 3) translocation (2% of cases).

Down syndrome is also the most studied syndrome from various points of view, especially in recent decades. A good amount of data is available on this subject, but this does not mean that all aspects of the syndrome, its development and related pathologies and problems have been fully identified. Therefore, there is often a tendency to consider this syndrome, from the point of view of language or other psychological and psychobiological functions, as prototypical of moderate to severe mental retardation. This is excessive, dangerous and most likely inaccurate. The evidence we are beginning to gather about other genetic syndromes with ID suggests greater variability and a certain degree of syndromic specificity [6,11].

Williams Syndrome

This syndrome originates from a rare metabolic disorder (approximately one in 20,000 births, with a higher incidence in males, 63%, than in females). The aetiological mechanism responsible for Williams syndrome is an abnormality on chromosome 7. The psychological profile of individuals is distinctive and most often includes: 1) a marked dissociation between language and general cognitive abilities; 2) the existence of a severe spatial cognition deficit; and 3) significant motor problems, especially 'fine' motor skills.

Individuals with Williams syndrome more frequently present a

dual motor characteristic. On the one hand, there is a weakness in visual perception, which causes these children and adolescents to have great difficulty integrating the different parts of a visual whole into a coherent and functional whole. This type of problem is evident in drawings and graphic representations. For example, they have been observed to be poor at representing complex objects such as bicycles. Their drawings show a 'scattering' of the 'parts' that make up the object, which is 'assembled' without functional integration, unlike individuals with Down syndrome, who often draw poorly. Still, the object is 'intact' and therefore easily recognisable. The other aspect concerns a lesser impairment of the language area, at least regarding the formal elements of language functions (articulation, grammar), which are partially preserved in this syndrome.

On the other hand, communication and the pragmatic (i.e. social) functioning of language (e.g. taking the interlocutor into account, conversing correctly and effectively according to the cultural and linguistic norms in force in the community) are problematic. There is therefore a clear contrast between the language problems of individuals with Williams syndrome and those with Down syndrome (Table 1). The latter more often have significant problems with articulation and morphosyntax. However, their communicative and pragmatic functioning is usually adequate, albeit with reduced formal means.

Fragile X Syndrome

Individuals with Fragile X syndrome have a null mutation (meaning that the gene in question is no longer able to function as expected) of the FMR-1 gene (at position q27 on the X chromosome), resulting in significantly reduced levels of DNA (deoxyribonucleic acid) protein. Many cases of Fragile X remain unidentified, making it difficult to establish the exact incidence of the syndrome. Eighty per cent of affected males have moderate mental retardation; the others have normal intelligence. About one-third of female carriers are affected by a variant of Fragile X syndrome that results in learning difficulties. Some of these women have mild mental retardation. The others are normal, but can pass on the genetic problem to their children.

Individuals with Fragile X syndrome have difficulties with verbal and visual memory, attention deficits, hyperactivity, and a tendency to impulsiveness. Males, in particular, have considerable challenges with controlling the rhythm of speech. Vocal problems, dysrhythmia, echolalia and reduced speech intelligibility are also observed. These individuals may exhibit verbal perseveration, and their language may be pragmatically inappropriate.

Autism Spectrum Disorder

Autism spectrum disorders (ASD) represent a vast diagnostic category that includes a wide range of ages and levels of functioning. What the different individuals have in common are deficits in social communication, social interaction and restricted/repetitive patterns of behaviour, interests or activities [12], as well as other non-social factors, such as stereotypies, the occurrence of intellectual disability and sensory problems, which play an essential role in the functioning profiles of individuals with ASD (Cardillo, 2018). According to recent estimates, the disorder can vary from one case in every 36 children, as in the United States (Centers for Disease Control and Prevention – CDC; 2020), to one in every 77 in Italy (Istituto Superiore di Sanità – ISS; 2021), or even lower or higher incidences in other countries.

Comorbidity between autism and intellectual disability is prevalent, with approximately 50-70% of people with autism also having an intellectual disability. This means the two disorders often coexist and can influence each other's clinical picture. The presence of intellectual disability can affect the manifestation of autistic symptoms, making recognition and diagnosis more difficult. It can also have a greater impact on mental health challenges, such as an increased risk of epilepsy, gastrointestinal disorders and sleep disorders. In addition, people with ASD may have greater difficulties in school learning, communication and the development of social skills.

A careful reading of these data provides many significant insights into the cognitive factors involved in learning.

Cognitive Factors and the Human Information Processing Model

Cognitive processes can be defined as the mental representations and processes (perception, attention, memory, thought) that allow individuals to perceive and process the information underlying behaviour and to know the world. This approach began in the 1960s and has been most clearly expressed in the Human Information Processing (HIP) paradigm, which considers human beings as 'information processors' [13].

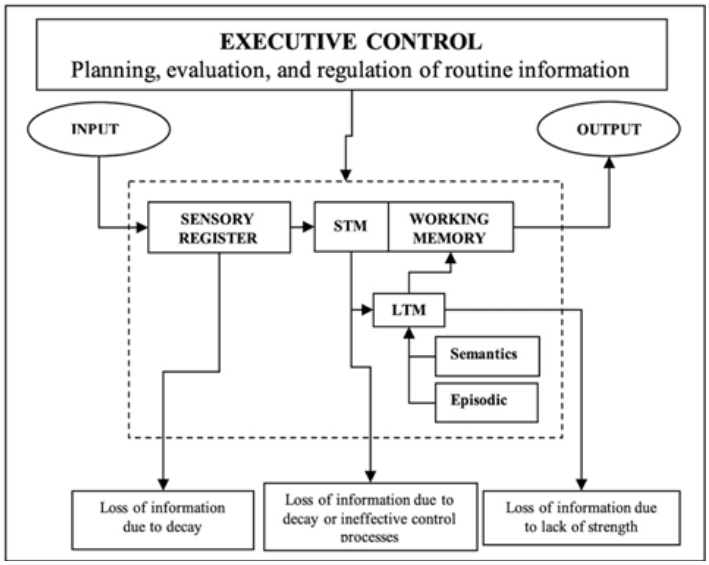

Figure 1: In the HIP model, information is processed and stored in three stages: 1) sensory register (stimuli can be 'internal' or 'external' to the organism), 2) Short-Term Memory (STM), also called Working Memory (stimuli 'exit' the system in the form of thoughts or behaviours), and 3) Long-Term Memory (LTM), which includes several subsystems, including 'semantic' and 'episodic' memory [8].

As can be seen in Figure 1, information processing can take place at different levels and can be either sequential (*serial processing*) or multiple (*parallel processing*) [14,15]. Furthermore, it can occur both in the person (recalling knowledge and linking it with knowledge from the external environment) and between the individual and the environment (reading, observing, etc.).

In this last stage, the individual receives information from the outside world through the sense organs, each of which is connected to a sensory register: the data is stored for a short time (about one in the register corresponding to the sensory stimulus (visual/visual, acoustic/acoustic, etc.).

In the sensory register, information can undergo two types of processing: it is either ignored and therefore discarded, or, if recognised as relevant, it is recoded and passes on to subsequent stages of processing. Recognition occurs by comparing the information with knowledge stored in long-term memory. These sensory memories are best understood as part of primary processing. Both sight and hearing seem to have another temporary store, which temporarily stores the term visual and auditory memory, in which the memory trace lasts a few seconds (obviously, in addition to these stores, we also have long-term visual and auditory memory).

This allows sensory information to be integrated with information from other sources through the limited-capacity operations of the working memory system. This information is also sent to the long-term memory system, which, although based primarily on encoding in terms of meaning, can also store sensory characteristics such as those involved in memory for faces, voices, tones, etc. These characteristics are probably stored as part of a multidimensional mnemonic trace. The information is then sent to the Short-Term Memory (STM) [16].

In the STM, information is stored for a slightly extended period (tens of seconds): a more extended system of storing only a few pieces of information at a time and only for limited periods. A typical role of short-term memory is sequential, consisting of maintaining the order in which information is presented to avoid omissions, anticipations, postponements, etc., which would distort the message or make the output inaccurate.

Therefore, the interest of the cognitive component does not lie in its isolated characteristics, but in their relationship with the entire system. It is not very interesting to see how we can remember a group of numbers for a few seconds. Still, it is essential to analyse how this ability functions in our cognitive activity. In STM, we can find the working function and the immediate recall process discussed above. The term 'working memory' refers to those parts of the human memory system that temporarily retain information and operate on it to perform a wide range of mental activities. Reading, writing, speaking, hanging a picture, etc., are, in fact, activities that require us to operate on content that cannot all be present at a given moment, but which is partly retained in the mind due to a temporary memory system with limited capacity, for the time necessary to perform the mental operations required by the situation. Based on these considerations, Baddeley [17] proposed a multicomponent working memory consisting of a central executive control system and two other dependent subsystems.

The first subsystem, the *articulatory loop*, is, according to Baddeley, responsible for maintaining verbal information and consists of a passive store and an active articulatory process. This simple model can explain several factors that influence short-term memory, including acoustic similarity, word length, unattended speech, articulatory suppression, etc. In addition, the articulatory loop is essential in learning to read, understanding language and acquiring vocabulary. However, it should be borne in mind that working memory is constantly involved in these skills to retain certain information: readers must remember the information they have just read to understand better what follows, those learning to read must memorise sequences of phonemes to blend them, those writing must remember previous parts of the text to understand it and subsequent parts that have not yet been written (for example, if we are dictated a sentence consisting of five words, while writing the first one we must remember all the others), those doing calculations must retain information relating to rules and operations already performed [17].

The second subsystem identified by Baddeley is the *visuospatial sketchpad*, a system responsible for the preparation and manipulation of visual images, but not for the effect of 'imaginability' in verbal long-term memory. It is probably a multifaceted system with both visual and spatial dimensions. Several studies [18] have highlighted that the visual system consists of two components, one linked to the processing of complex stimuli and the identification of 'what' (*what system*). At the same time, the other is involved in the spatial localisation of stimuli and conveys information about 'where' (*where system*). These considerations suggest that imagination has related but separable visual and spatial components [19].

Most research on working memory has focused on subsidiary systems, as these allow us to tackle problems that are more manageable than those posed by the central executmore manageable problemsthird component has the function of selecting strategies, managing available attentional resources, integrating information from different sources, and coordinating the execution of various activities when they are performed simultaneously, to achieve the best performance. In a sense, therefore, the central executive functions more as an attentional system than as a memory store.

Finally, information passes into Long-Term Memory (LTM), which, unlike short-term memory, is permanent and has unlimited capacity. Here, information is stored and, when necessary, retrieved through a search process. A long-term data repository is where everything an individual knows is stored: names, experiences, knowledge, etc. A fundamental feature of memory is that strategy is a method of achieving a goal. It involves a more or less controlled attempt to adapt cognitive processes to the requirements of a task to accomplish a goal. Furthermore, strategies are modifiable and can be made more effective. Ability to perform a task can be hindered by three types of problems: limited capacity, not knowing how to

use the appropriate strategy, and the inefficiency of the methods used [20].

Using strategies as a basic element of knowledge has two fundamental implications at the psycho-pedagogical level. The first implication is the dynamic nature of knowledge acquisition, which proceeds through the modification and reorganisation of structures that process experience and are influenced by it. The second implication concerns the continuity between what is acquired and what is new, between what is remembered and what is learned; on the other hand, if knowing is always a process of constructing information based on accumulated knowledge, learning is never repetitive. Some authors [21,22] have considered learning as the result of the interaction between different cognitive processes and between these and metacognitive processes; that is, they have developed interactive models of learning that allow the relationship between the following factors to be analysed:

- ***the individual characteristics of the learner*:** their specific cognitive and metacognitive abilities, their strategic knowledge, their cognitive styles, their motivations, attributions and expectations, etc.;
- ***the learning* activities:** the cognitive processes (attention, comprehension, thinking, etc.) implemented and the strategies used by the learner in that particular task;
- the characteristics of the material to be learned: the texts, the methods and order of presentation, the presence of questions, diagrams, etc.;
- **the characteristics of the *quality of education*:** the teacher's methodological choices and ability to communicate with students;
- **the criterion task (test, always chosen by the teacher to assess learning):** questionnaire, multiple-choice questions, oral examination, etc. Several factors influence the result of a criterion task: the students' knowledge, expectations of themselves, and beliefs. Several factors influence the result of a criterion task, including the type of material used for learning and the instruction received.

All these factors can be attributed to a type of school problem related to the learning process, which is defined by the generic term 'learning difficulties'.

Learning Difficulties in Intellectual Disabilities

Learning difficulties are an umbrella term that covers a diverse range of problems in cognitive development and school learning. They can be defined as the failure to achieve certain relevant learning criteria.

According to some, learning difficulties are problems translated into school education. According to this logic, difficulties can be traced back to all those domains for which a given educational context places demands that specific individuals cannot meet. In practice, reference has mainly been made to problems with reading, writing and arithmetic, but – as mentioned above – it is to be expected that attention will soon shift more significantly to other areas of school learning.

***Reading disorders*.** Among the problems related to reading, learning disorders are probably the best known and most studied. However, they have often been (erroneously) included in different clinical pictures such as *developmental dyslexia* and *psychomotor instability*. Reading decoding skills, which can be assessed by estimating accuracy and fluency in reading aloud, and comprehension skills, which can be assessed through questionnaires based on the subject's reading, are essentially independent, a finding that is confirmed in the international literature but appears to be particularly true an, whose phonological regularity allows people to read even without understanding what they are reading [1,8].

***Writing disorders*.** Johnson and Myklebust [23] were among the first scholars to characterise writing disorders systematically. In this characterisation, for problems with graphism, which are more related to visuosystematically characterisevelopment, they recognised the difficulties involved in reproducing a graphic sign from memory (e.g., remembering how to write a letter) or in copying it (e.g., copying a letter by looking at a model). These problems, linked to disorders traditionally classified as 'praxic', cause difficulties in writing because the child has poor control over the graphic stroke and the space on the page.

As regards 'written discourse', on the other hadifficultiere are numerous problems of analysis linked to the complexity of the process [24]: the transition from the interactive characteristics of oral production (language) to the characteristics of a 'graphic' system (writing), which requires the management of cognitive processes at various levels (lexical access, spelling, grammar, planning, revision, taking the perspective of the potential reader, etc.).

Calculation disorders. Studies by Rourke and Strong [25] and Badian [26] describe and classify developmental dyscalculia, hypothesising its basis in cognitive and neuropsychological structures. Rather than these disorder classifications, recent research refers to neuropsychological models of numerical knowledge and calculation. Processing developed mainly from studying adult subjects [27-29], highlighting its characteristics in children.

Research by Temple [30] and Macaruso and Sokol [31], focusing on the mechanisms involved in calculation in developing subjects and drawing inspiration from McCloskey's modular neuropsychological model [28], have shown that the mental representation of numerical knowledge, in addition to being independent of other cognitive systems, is structured in three functionally distinct modules.

According to this model, the calculation system takes a representation as input and then manipulates it through the functioning of three components: *digit dyslexia*, in which lexical mechanisms are impaired, while syntactic mechanisms are

adequately developed; *procedural dyscalculia*, characterised by difficulty in acquiring calculation procedures in the absence of errors related to numerical processing; *dyscalculia for 'arithmetic facts'*, characterised by problems in acquiring multiplication tables, basic operations, etc. within the calculation system.

Therefore, processing a number initially involves its conceptual or semantic representation, through which all the elements that make up the number are identified, specifying for each of them the information concerning quantity and order of magnitude. This information regulates the lexicon of numbers and is closely interdependent with the syntax that regulates the numbers themselves (positional value of digits).

Language disorders. In typically developing children, the simultaneous development of different skills is probably triggered by maturation and differentiation processes, the integration of which allows increasingly complex mental functions to emerge. This integration may not occur in pathological conditions, leading to a dissociation between language's cognitive or semantic-conceptual aspects and its formal aspects. Examples of conceptual-formal dissociation are represented by some cases of psychosis and/or mental retardation in which the phonological and morphosyntactic aspects of language appear better preserved than the conceptual components. In contrast, formal-conceptual dissociation is typical of specific language disorders, congenital and acquired, which are characterised by a disorder in the acquisition or reacquisition of language despite adequate cognitive, affective and social development [32].

Primary language disorder is diagnosed when a child of normal intelligence with normal overall development presents a discrepancy in language processing relative to their chronological age. Most authors agree that a primary disorder is needed when a child speaks little or poorly between the ages of two and four, i.e., during the period that accelerates typical language development.

A secondary language disorder is diagnosed when an underlying condition, such as a neurological or medical condition, causes a delay in language development. An underlying condition, such as a neurological disorder or a medical condition, causes the delay in language development of various kinds, manifesting itself in different ways at different stages of development, such as in procedural memory management, motor control, phonological working memory, and executive functions.

The most productive approach to studying learning disorders in children with intellectual disabilities is to always consider two sets of problems in parallel. On the one hand, a series of basic cognitive disorders limits the quantity and quality of mental operations possible at each stage of development; on the other hand, the evolution of intellectual disability is a development of functions and dysfunctions that can integrate atypically, compete or dissociate. This also means that children with atypical development are always at risk of confusing and/or disabling reactions when faced with new learning: as if sometimes the child risks not learning little, but learning *too much*, and then not knowing *why and how* to use what they have learned.

Conclusions

The learning difficulties experienced by people with ID during their schooling can take various clinical forms, such as simple slowing down (delay), severe deficits (hypo-: in reading, arithmetic, language, etc.), as exceptional abilities (hyper-: in reading, arithmetic, language, etc.); all of these developments are possible but difficult to predict and assess if analysed solely in strictly intellectual terms. This heterogeneity requires the use of reliable parameters, such as the time taken to reach certain developmental milestones (developmental indicators), which help specialists to make an early diagnosis of the severity of the delay (diagnosis of 'specificity') and, at a later stage, to analyse the appearance of specific simple but significant indicators (such as syllables) that allow the evolution and mode of learning to be assessed.

Delayed diagnosis of learning disorders often prevents timely and appropriate care for children with intellectual disabilities, which risks hindering their adequate schooling, depending on the severity: in mild cognitive delay, treatment should be aimed at achieving better logical-linguistic integration; in moderate cognitive delay, treatment should be aimed at strengthening the link between practical and linguistic skills; while in severe mental retardation, treatment should be geared towards functional learning curricula (words that are ecologically meaningful and relevant to the person's existence, etc.).

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