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ORIGINAL ARTICLE

Study of the Specific Language Impairment in a three-generation family

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SLI genetics;
FOXP2 mutation;
Apraxia of speech;
Oral-motor dyspraxia

Abstract
Purpose: Three members of a family, one of each generation, are studied in order to obtain their SLI language profile, together with motor and cognitive data. Additional information on the absence of FOXP2 mutations is also provided.
Method: The language profile is twofold: natural conversation and language tests, and an evaluation of their cognitive abilities, oral-motor praxis, and laterality.
Results: Cognitive abilities (short term and procedural working memory, perception, conceptual and coherence strategies) are not at the average level. General oral fine mobility (not the speech apraxia), fluency and auditory phonetic discrimination are impaired at different degrees. The language phenotype exhibits lexical as well as syntactic processing difficulties as the main impairments. The language competence of the adult members is simple but sufficient for everyday communication.
Conclusions: The long-standing language competence results of SLI show an adaptation in terms of simplicity, high frequency strategies, and pragmatic resources. Language profile, sensory-motor abilities and cognition favour a non-specific approach to the language acquisition impairment.

PALABRAS CLAVE
Trastorno específico del lenguaje (TEL);
Trastorno del lenguaje (TL);
Cognición en el TEL;

Estudio del trastorno específico del lenguaje en tres generaciones de una familia

Resumen
Objetivo: Estudiar tres miembros de una familia con Trastorno Específico del Lenguaje (TEL), uno de cada generación, para obtener su perfil de lenguaje, junto a otros datos de tipo motriz y cognitivo. Se aporta información adicional sobre la ausencia de mutaciones en el gen FOXP2.

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Memoria de trabajo procedimental en el TEL;
Bases genéticas del TEL;
Mutación del gen *FOXP2*;
Apraxia del habla;
Dispraxia oromotora

Método: El perfil del lenguaje que se presenta es doble: Los datos provienen de conversaciones naturales y de tests de lenguaje. También son evaluadas sus habilidades cognitivas, sus praxias oro- motrices y su lateralidad.
Resultados: Las habilidades cognitivas de las memorias a corto termino y procedimental de trabajo, la percepción, la conceptuación y las estrategias de construcción de coherencia (texto), no alcanzan un nivel de normalidad. La movilidad fina oral general (no la apraxia de habla), la fluidez, y la fonética auditiva son deficientes en grados diversos. El fenotipo del lenguaje manifiesta sus máximas dificultades de procesamiento en los niveles léxico y sintáctico. Los adultos tienen un lenguaje coloquial muy simple pero normal.
Conclusiones: La competencia lingüística de los miembros mayores muestra una buena adaptación, resaltando sus estrategias de simplicidad, usos de alta frecuencia y buena pragmática comunicativa. El perfil del lenguaje, las habilidades sensorio – motrices y las cognitivas favorecen una aproximación no especifica al TEL.

Introduction

It is widely accepted that Specific Language Impairment (SLI) refers to a neurodevelopmental disorder which impairs normal language competence and use, in a context of an adequate education and environment. The absence of other causal explanations such as mental or sensory deficits is a necessary condition for a SLI diagnosis (Bishop & Leonard, 2000; Leonard, 2014). However, there is evidence that SLI may be associated with either a genetic background – 24% of cases (Bishop & Edmunsen, 1986; Marcus & Fisher, 2003) – or/and mild deficits in other non-linguistic cognitive abilities (Li & Bartlett, 2012). Some authors (Law, Boyle, Harris, Harkness, & Nye, 2000) suggest that the disorder affects from 5% to 8% of school age children. Some others, with different diagnostic criteria (Serra, 2002) suggest that it affects 0.3%.[1]

In this paper, a Spanish family will be under study because they exhibit an autosomal-dominant monogenic transmission (see Fig. 1), similar to the disorder described in the well-known English KE family.[2] In addition to that family distribution, in the first interviews, other mild perceptual – cognitive – motor were suspected.

In the 1990s an English family, the KE family, received a lot of attention because of the high number of individuals affected by a severe language problem (16/25). Mutations in the FOXP2 gene were found in their phenotype and according to the gathered evidence were considered as causally related to SLI (Fisher, Lai, & Monaco, 2003). Finally, after re-examination of symptoms, their phenotype was considered as a severe developmental verbal dyspraxia, impaired use of morpho-syntactic rules and severe extra-linguistic oro-facial dyspraxia (Watkins, Dronkers, & Vargha-Khadem, 2002). Later on, other studies have not found any FOXP2 mutation in SLI persons, avoiding the simplistic model of ''the gene of language''. Nonetheless, today, the genetic mutations, probably many of them, are considered important contributing factors to language and other developmental difficulties, and are the subject of many research projects (see for a recent review of the question, Fisher & Vernes, 2015).

Purpose

The aim of this study is to provide a comprehensive phenotype of SLI for a young subject and for two adults of their language competence, in natural as well as in test situations. Other abilities, including sensory, motor and cognitive evaluations, have also been measured. Special attention was paid to oro–motor abilities and verbal praxia[3] as possible comorbidity.

[1] Our strict criteria for SLI diagnosis require the presence of deviant (categorical) errors, like obligatory nuclei omissions, after one year of help or therapy in children of more than four years. If only functional errors are present (article omissions, agreement errors that are semantically and pragmatically irrelevant, etc.), the subject is diagnosed as language delayed (Serra, 2002).

[2] Puede consultarse Wikipedia para su historia y visitar you tube para ver videos de uno de los miembros afectados: https://www.youtube.com/watch?v=Fg2rLOkoL9Q.

[3] Verbal praxia correspond to a fluent motor articulation of learned assemblies of connected speech. Other specific, but non-verbal praxia, are seen in non symbolic sequences of sounds like animal imitation, whistelling, blowing, balloon inflating, etc. Imitation of non-sense/non-possible words would be on the border between the two types of praxia (Varley, 2011). Similar to the reading impairment case, here we should make the distinction between ''Child apraxia of speech'' for the learning difficulties and low competence, in order to distinguish them from their loss. In the case of Apraxia of speech, the child knows what he or she wants to say, but his/her motor brain has difficulties in coordinating the muscle movements necessary to utter those words.