Brain & Language 159 (2016) 102–108

Contents lists available at ScienceDirect

Brain & Language

journal homepage: www.elsevier.com/locate/b&l

Intrafamilial phenotypic variability of Specific Language Impairment

Lisa Bartha-Doering [a,*], Sabrina Regele [b], Dirk Deuster [b], Rainer Seidl [a], Nadja Bogdanova [c], Albrecht Röpke [c], Peter Wieacker [c], Antoinette am Zehnhoff-Dinnesen [b]

[a] Department of Pediatrics and Adolescent Medicine, Medical University Vienna, Waehringer Guertel 18-20, 1090 Vienna, Austria
[b] Department of Phoniatrics and Pedaudiology, University Hospital of Muenster, Kardinal-von-Galen Ring 10, 48149 Muenster, Germany
[c] Institute of Human Genetics, University Hospital Muenster, Vesaliusweg 12-14, 48149 Muenster, Germany

ARTICLE INFO

Article history:
Received 11 February 2016
Revised 8 June 2016
Accepted 15 June 2016
Available online 4 July 2016

ABSTRACT

We investigated language functions in 32 members of a four generation family with several members affected by Specific Language Impairment with an extensive language test battery in order to determine the prevalence, overlap, and homogeneity of linguistic deficits within one pedigree. In sum, one fourth of all family members tested fulfilled the criteria of Specific Language Impairment. Despite of some similarities in language abilities, different combinations of language deficits were observed, and individual language profiles varied substantially. Thus, though there is a high prevalence of language deficits in this family which raises the likelihood of a genetic origin of these deficits, and though all affected study participants displayed selective linguistic deficits with normal non-verbal functioning, language testing showed considerable variance in overlap and homogeneity of linguistic deficits. Thus, even in one genetic population, an underlying linguistic disorder manifests itself in different language abilities to a variant degree.

© 2016 Elsevier Inc. All rights reserved.

1. Introduction

Specific Language Impairment (SLI) is defined as a prominent delay in language development in the absence of nonverbal cognitive deficits, neurological damage, psychiatric disease, or peripheral hearing loss (Leonard, 1998). SLI affects up to 7% of children attending kindergarten and is therefore recognized as a frequent neurodevelopmental disorder in children (Tomblin et al., 1997). While approximately half of the children are able to catch up their oral language deficits until school start, the other half of children continues to show deficits in morphology, syntax, vocabulary, and written language acquisition (Conti-Ramsden, St Clair, Pickles, & Durkin, 2012; Critten, Connelly, Dockrell, & Walter, 2014). SLI is associated with poor academic outcomes and influences the professional development and psychosocial aspects of life (Conti-Ramsden & Botting, 2008; Johnson, Beitchman, & Brownlie, 2010; Wadman, Durkin, & Conti-Ramsden, 2008; Whitehouse, Watt, Line, & Bishop, 2009). SLI thus represents a major public health problem.

SLI is a heterogeneous disorder. Subjects with SLI display differences in the severity of the disorder, the areas of language affected by the disorder, and the stability of their language profile over time (Conti-Ramsden & Botting, 1999). This variation has led to the definition of different SLI subgroups (Bishop, 2004; Conti-Ramsden, Crutchley, & Botting, 1997). Furthermore, SLI often overlaps with other cognitive impairments such as semantic-pragmatic disorder, the autistic spectrum, or attention deficit hyperactivity disorder (Bishop, 2003; Tirosh & Cohen, 1998).

The heterogeneity of SLI has been attributed to its complex aetiology, where both genetic and environmental factors are likely to contribute (Conti-Ramsden, Falcaro, Simkin, & Pickles, 2007).

SLI aggregates in families. In studies collecting family history information with questionnaires, approximately 20–60% of families with a member affected by SLI report language impairments in at least one other immediate family member, compared to 18% in families without a history in SLI (Bishop & Edmundson, 1986; Tallal, Ross, & Curtiss, 1989; for a review of 18 studies, see Stromswold, 1998). Studies using direct language testing reported similar rates of impairment. Tomblin and Buckwalter (1994) found that 42% of individuals diagnosed with SLI had at least one family member with a speech or language problem, and Tallal et al. (2001) reported that 52% of affected children had a first degree relative with language impairment, compared to 15.4% of control children. Choudhury and Benasich (2003) described that children born into families with a positive history of SLI scored significantly lower

* Corresponding author.
E-mail addresses: elisabeth.bartha-doering@meduniwien.ac.at (L. Bartha-Doering), sabrina.regele@uni-muenster.de (S. Regele), d.deuster@uni-muenster.de (D. Deuster), rainer.seidl@meduniwien.ac.at (R. Seidl), bogdano@uni-muenster.de (N. Bogdanova), albrecht.roepke@ukmuenster.de (A. Röpke), wieacker@uni-muenster.de (P. Wieacker), am.zehnhoff@uni-muenster.de (A. am Zehnhoff-Dinnesen).

http://dx.doi.org/10.1016/j.bandl.2016.06.005
0093-934X/© 2016 Elsevier Inc. All rights reserved.

in language measures than children without family history of SLI, and were more likely to fall below the 16th percentile (28%) than control children (7%). In addition to family aggregation studies, twin studies have shown that SLI has a highly heritable component. A meta-analysis of twin studies exhibits overall concordances of 84% for monozygotic twins and 50% for dizygotic twins (Stromswold, 2001).

Genetic studies have identified different loci of interest and a number of candidate genes that have been associated with quantitative measures of language skills (Reader, Covill, Nudel, & Newbury, 2014; Villanueva et al., 2015). However, the understanding of the specific genetic mechanisms underlying SLI has proved challenging, and contributions of these various genetic effects seem to be complex. Environmental factors potentially hampering language development include parental language input, parents' educational level, and income (Bishop, Adams, & Rosen, 2006).

Thus, many factors seem to contribute to the heterogeneous picture of SLI. We were therefore interested in the degree of homogeneity of language deficits in a group of patients with similar environment, the same cultural background, and foremost, within a group of genetically related individuals. We therefore investigated all available family members of a four generation family with several members affected by SLI with an extensive cognitive test battery in order to determine the prevalence, overlap, and homogeneity of linguistic deficits within this family.

2. Methods

2.1. Participants

We studied one pedigree originating from Germany, where four siblings and their mother attracted our attention because of their severe SLI. We then contacted all living family members of the mother (n = 43) both via letter and telephone. Inclusion criteria were an age between 4 and 80 years, normal hearing, measured with a pure tone audiogram, and native, monolingual German speakers. Subjects with neurological or neurometabolic diseases were excluded from this study. Written informed consents were obtained from all participants, or in the case of minors, from the parents. The study was approved by the Ethics Committee of the Medical University Muenster in accordance with the Helsinki Declaration of 1975.

2.2. Procedure

An extensive neuropsychological and neurolinguistic test battery was performed in all subjects. The psychometric assessment tapped various language domains including (a) verbal-auditory short-term and verbal working memory, (b) reading aloud and writing, (c) language comprehension, (d) morphology and syntax; and (e) word fluency. In addition, the nonverbal IQ was measured from the age of 6 years on. Due to the large age-range of participants, different language tests in different age-groups were applied in order to be able to compare results to standardized norms. For inter-subject comparisons, raw scores of cognitive tests were transformed into age-adjusted percentiles for each cognitive test. For reading, writing, and comprehension, adult normative data unfortunately only provide cut-offs.

The span of immediate verbal recall was measured by digit span tasks of Kaufmann Assessment Battery for Children (Melchers & Preuß, 2009), the Hamburg-Wechsler-Intelligenztest für Kinder IV (Petermann & Petermann, 2008), and the Hamburg-Wechsler-Intelligenztest für Erwachsene (Tewes, 1994), respectively. These tests consist of pairs of random number sequences of increasing length read by the examiner at a rate of one per second. The Digits Forwards test, where the participant has to repeat the presented number sequences, is thought to measure auditory-verbal short-term memory. The Digits Backwards test, evaluated from the age of six on, requires the participant to repeat sequences of digits in the reverse order, and is interpreted as a measure of verbal-auditory working memory.

Writing abilities were evaluated with writing to dictation. For the younger participants from 8 to 10 years, cloze texts of the Salzburger Lese- und Rechtschreibtest SLRT II (Moll & Landerl, 2010) were used. Adolescents with ages between 10 and 15 years were tested with the Hamburger Schreibprobe (May, 2002) which requires the individual to write single words and sentences. In adults, writing to dictation of 30 words with regular and irregular spelling was tested with the subtest 21 of Lexikon Modellorientiert LEMO (De Bleser, Cholewa, Stadie, & Tabatabaie, 2011). For qualitative analysis, spelling errors were classified into phonological and orthographic errors. Errors were defined as phonological errors when they did not have an adequate phoneme-to-grapheme correspondence in German, thus violated the phonological principle, for example *lobstser* instead of *lobster*. These errors typically involve the substitution of wrong sounds, insertion of phonemes, or deletion of phonemes. Errors were defined as orthographic errors when they preserved the phonology of the word, but were orthographically incorrect. These errors most often occur in words whose spellings cannot be derived on the basis from phonology but have to be memorized, including loan words with deviant phoneme-grapheme correspondences and words with phonemes that can be spelled in different ways.

Reading skills were assessed from the age of 6;6 on using tests of reading fluency (SLRT II), reading speed and comprehension (Lesegeschwindigkeits- und Verständnistest LGVT, Schneider, Schlagmüller, & Ennemoser, 2007), and reading aloud of pseudowords and nonwords (LEMO subtest 17). In both SLRT II and LGVT, more than one percentile rank was obtained. To be able to compare the output with the adults' test from LEMO, a composite score was calculated.

Language comprehension was evaluated using the Token Test for Children TTFC-2 (McGhee, Ehrler, & DiSimoni, 2007) and the Token Test of the Aachener Aphasietest (Huber, Poeck, Weniger, & Willmes, 1983), respectively. It consists of 20 tokens varying in colour, size and shape and requires response to commands by manipulating the tokens. The commands are arranged in different blocks of increasing linguistic and memory demands.

Competence of morphological rules was tested with the Heidelberger Sprachentwicklungstest HSET (Grimm & Schöler, 1991), where production of noun plurals, derivational morphemes, and derivations from nouns to adjectives in regular, comparative, and superlative form were evaluated.

Verbal fluency was evaluated by requiring the participant to name as many words as possible starting with a specific letter in 2 min (Regensburger Wortflüssigkeitstest RWT, Aschenbrenner, Tucha, & Lange, 2001).

Nonverbal IQ was tested using the Standard Raven's Progressive Matrices (Raven, 1981). This test is made of 60 multiple choice questions, listed in order of difficulty, and the subject is asked to identify the missing element that completes a pattern.

An impairment in a cognitive domain was diagnosed when test performance lay below the defined cut-off or below the 10th percentile (SD −1.25), respectively, a cut-off commonly used in clinical practice (Conti-Ramsden et al., 2007). Following the definitions of Tomblin et al. (1997) and Leonard (1998), SLI was defined if the participant met the following criteria: normal hearing, nonverbal IQ $\geqslant$ 85, and performance below the cut-off on at least two language domains.

In addition, at study induction, adult participants and parents of participating children, respectively, were asked to complete a

Download English Version:

https://daneshyari.com/en/article/7283893

Download Persian Version:

https://daneshyari.com/article/7283893

Daneshyari.com