Journal of Communication Disorders 65 (2017) 22–34

Contents lists available at ScienceDirect

Journal of Communication Disorders

Signaling of noncomprehension in communication breakdowns in fragile X syndrome, Down syndrome, and autism spectrum disorder

Gary E. Martin[a,*], Jamie Barstein[b], Jane Hornickel[b], Sara Matherly[c], Genna Durante[c], Molly Losh[b]

[a] Department of Communication Sciences and Disorders, St. John's University, Staten Island, NY, USA
[b] Roxelyn and Richard Pepper Department of Communication Sciences and Disorders, Northwestern University, Evanston, IL, USA
[c] Frank Porter Graham Child Development Institute, University of North Carolina at Chapel Hill, Chapel Hill, NC, USA

ARTICLE INFO

Article history:
Received 8 February 2016
Received in revised form 13 January 2017
Accepted 19 January 2017
Available online 26 January 2017

Keywords:
Noncomprehension
Communication breakdown
Pragmatic language
Fragile X syndrome
Autism spectrum disorder
Down syndrome

ABSTRACT

The ability to indicate a failure to understand a message is a critical pragmatic (social) language skill for managing communication breakdowns and supporting successful communicative exchanges. The current study examined the ability to signal noncomprehension across different types of confusing message conditions in children and adolescents with fragile X syndrome (FXS), Down syndrome (DS), autism spectrum disorder (ASD), and typical development (TD). Controlling for nonverbal mental age and receptive vocabulary skills, youth with comorbid FXS and ASD and those with DS were less likely than TD controls to signal noncomprehension of confusing messages. Youth with FXS without ASD and those with idiopathic ASD did not differ from controls. No sex differences were detected in any group. Findings contribute to current knowledge of pragmatic profiles in different forms of genetically-based neurodevelopmental disorders associated with intellectual disability, and the role of sex in the expression of such profiles.

Learning outcomes: Upon completion of this article, readers will have learned about: (1) the social-communicative profiles of youth with FXS, DS, and ASD, (2) the importance of signaling noncomprehension in response to a confusing message, and (3) the similarities and differences in noncomprehension signaling in youth with FXS (with and without ASD), DS, idiopathic ASD, and TD.

1. Introduction

Pragmatic, or social, language skills are often impacted, to varying degrees, in individuals with intellectual and developmental disabilities such as fragile X syndrome (FXS), Down syndrome (DS), or autism spectrum disorder (ASD) (Abbeduto, Brady, & Kover, 2007; Rice, Warren, & Betz, 2005; Tager-Flusberg, Edelson, & Luyster, 2011). Pragmatics is a multifaceted domain comprised of a complex array of component skills, such as speech acts, topic maintenance, turn-taking, and repair of communication breakdowns. One critical skill for repairing breakdowns in communication is a listener's ability to indicate when a speaker's message is not understood, or to signal noncomprehension of a confusing or unclear message.

* Corresponding author at: St. John's University, DaSilva Academic Center, Room 348, Staten Island, NY 10301, USA.
E-mail address: marting@stjohns.edu (G.E. Martin).

http://dx.doi.org/10.1016/j.jcomdis.2017.01.003
0021-9924/

Typically developing children as young as two and a half years can signal noncomprehension of very noticeable incompatible messages (e.g., asking "Where is it?" when told to do something with an unavailable referent), whereas the skill for managing more difficult messages, such as those containing ambiguity or unfamiliar vocabulary items, continues to develop into the early school-age years (Flavell, Speer, Green, & August, 1981; Lempers & Elrod, 1983; Revelle, Wellman, & Karabenick, 1985).

The failure to indicate misunderstanding of a spoken message is likely to be particularly detrimental for individuals with neurodevelopmental disabilities, who may face communication breakdowns more frequently due to structural language and cognitive deficits or problems understanding and intuiting others' intentions. The current study compares noncomprehension signaling across three genetically-based neurodevelopmental disabilities–FXS, DS, and ASD–in order to determine whether any syndrome-specific differences exist that would inform general knowledge as well as the design of tailored language interventions. In the case of FXS and DS, we also examine sex differences (data were not available from girls with idiopathic ASD). Sex differences in pragmatic language have been documented in typically developing children (Austin, Salehi, & Leffler, 1987; Cook, Fritz, McCornack, & Visperas, 1985; Kothari, Skuse, Wakefield, & Micali, 2013; Leaper, 1991; Sigelman & Holtz, 2013). However, girls with neurodevelopmental disabilities tend to be underrepresented in research, and determining how phenotypes in genetic conditions may be differentially expressed in males and females has important implications for understanding underlying pathophysiology and informing differential assessment and intervention practices (Messinger et al., 2015; Rinehart, Cornish, & Tonge, 2011; Thompson, Caruso, & Ellerbeck, 2003). Finally, we include both males and females with FXS with and without ASD, along with males with idiopathic ASD, to examine the impact of ASD symptomatology on noncomprehension signaling within and across groups. Below we briefly review the literature on pragmatic skills in FXS, DS, and ASD.

1.1. Fragile X syndrome

FXS is the most common heritable cause of intellectual disability (Dykens, Hodapp, & Finucane, 2000; Hagerman & Hagerman, 2002). In FXS, methylation (turning off) of the Fragile X Mental Retardation-1 gene (*FMR1*) on the X chromosome leads to underproduction of the Fragile X Mental Retardation Protein (FMRP), a protein believed to be critical for normal cognitive functioning (Devys, Lutz, Rouyer, Bellocq, & Mandel, 1993; Jin & Warren, 2003; Patel, Loerwald, Huber, & Gibson, 2014). Because females possess two X chromosomes, females with FXS have one healthy FMRP-producing copy of *FMR1*. For this reason, females with FXS are often affected less severely than males (Hagerman & Hagerman, 2002; Loesch et al., 2002; Reiss & Dant, 2003). Whereas males with FXS generally have moderate to severe intellectual disability, females may display mild to moderate intellectual disability or perform in the normal range of intellectual functioning (Saldarriaga et al., 2014; Sterling & Abbeduto, 2012; Warren, Brady, Sterling, Fleming, & Marquis, 2010).

Pragmatic deficits that are beyond expectations for nonverbal cognitive ability are well documented in males with FXS, with characteristic features including noncontingent (tangential or off-topic) and perseverative (repetitive) language (Klusek, Martin, & Losh, 2014; Martin, Losh, Estigarribia, Sideris, & Roberts, 2013; Roberts et al., 2007; Sudhalter & Belser, 2001; Sudhalter, Cohen, Silverman, & Wolf-Schein, 1990). Far fewer studies have focused on the pragmatic language profile of females with FXS. In one study of parent-reported autistic behaviors in FXS, girls with FXS were rated as having more difficulty initiating and sustaining conversation than IQ-matched controls (Mazzocco, Kates, Baumgardner, Freund, & Reiss, 1997). Similarly, compared with age-matched peers, females with FXS took longer to initiate conversation (Lesniak-Karpiak, Mazzocco, & Ross, 2003) and asked fewer questions to maintain a topic (Mazzocco et al., 2006) in two other investigations. On a standardized test of expressive language abilities, Turkstra, Abbeduto, and Meulenbroek (2014) reported that adolescent females with FXS scored significantly lower overall than age-matched females with TD. In addition, 11 of the 20 girls with FXS had below-average scores on a pragmatic subtest. However, the TD group also scored significantly higher in nonverbal cognition and these differences were not controlled for in the language analysis. Because of the differences in general cognitive ability typically observed in boys and girls with FXS, very few studies have directly compared pragmatic language abilities in these groups. One exception is a study of repetitive language where males with FXS used more repetition of rote sayings and phrases (e.g., "that's a wrap") but did not differ from females in topic repetition (Murphy & Abbeduto, 2007).

Further complicating the pragmatic language profile in FXS is the common comorbidity of ASD, a developmental disorder defined in part by pragmatic language impairments. FXS is the most common known single-gene cause of ASD, with 40%–74% of males and 13%–45% of females with FXS meeting ASD criteria (Bailey, Raspa, Olmsted, & Holiday, 2008; Clifford et al., 2007; Hall, Lightbody, & Reiss, 2008; Kaufmann et al., 2004; Klusek, Losh, & Martin, 2014; Philofsky, Hepburn, Hayes, Hagerman, & Rogers, 2004; Rogers, Wehner, & Hagerman, 2001). Of note, IQ is generally lower in individuals with comorbid FXS and ASD (as is the case in many other monogenetic disorders showing strong overlap with ASD, e.g., Leblond et al., 2014) than in those with FXS only (Kaufmann et al., 2004; Philofsky et al., 2004; Rogers et al., 2001), raising the question of whether ASD in the context of FXS may simply reflect more severe cognitive deficits (see Abbeduto, McDuffie, & Thurman, 2014, for a review). However, in several studies that controlled for nonverbal cognitive ability, boys with comorbid FXS and ASD showed more pragmatic language deficits, including more noncontingent language and perseveration, than those without comorbid ASD (Klusek, Martin et al., 2014; Losh, Martin, Klusek, Hogan-Brown, & Sideris, 2012; Martin et al., 2013; Martin, Roberts, Helm-Estabrooks, Sideris, & Assal, 2012; Roberts et al., 2007). In studies using a semi-naturalistic conversational context or standardized test, boys with FXS and ASD also exhibited the same types of pragmatic language errors and severity of pragmatic impairment as boys with idiopathic ASD (Klusek, Martin et al., 2014; Losh et al., 2012).