Referential Choice in the Narratives of Italian Speakers with Klinefelter Syndrome

Mirta Vernice[1,*], Federica Clerici[2], Anna Cremante[2] and Annapia Verri[2]

[1]*Department of Psychology, University of Milano-Bicocca, Milano, Italy*

[2]*National Neurological Institute, IRCCS Mondino Foundation, Pavia, Italy*

Abstract: Klinefelter Syndrome (KS) is a genetic disorder characterized by an uneven neuro-linguistic profile. Whereas cognitive abilities appear to be within the normal range, KS patients often show poor linguistic abilities and language-based learning disorders. Although it has been proposed that KS can be considered a genetic model of language impairment, it is not yet well established whether speakers with KS are impaired in specific psycholinguistics aspects, such as reference production. The choice of an adequate referential expression (whether a full noun phrase, a null or overt pronoun, etc.) involves the use of memory mechanisms to represent the characters and actions involved, but also the ability to judge the attention and the knowledge of the hearer.

The present work focuses on KS speakers' ability to report a story based on a Sylvester and Tweety cartoon (Arnold *et al*., 2009). We examine the ability to choose an appropriate referent (overt pronouns *vs*. null pronoun *vs*. full noun phrase) made during a narrative by Italian adolescents and young adults with KS (n = 8) and age-matched typically developing controls. In addition, we administered to each participant a full battery of cognitive and linguistic tests.

Overall our results indicate that the correct use of referential expressions did not appear to be significantly predicted by the cognitive level of the speakers. Therefore the ability to choose an appropriate referential expression is preserved in KS whereas receptive vocabulary and comprehension skills are significantly lower as compared to controls.

Keywords: Klinefelter syndrome, reference choice, linguistic abilities.

INTRODUCTION

Klinefelter's syndrome (KS) is a genetic disorder characterized by the presence of one or more supernumerary X chromosomes, with an incidence of 1/500 male live birth. As many individuals with KS show no significant health problems, only one-third of them is usually diagnosed [1, 2]. Among the set of symptoms that might characterize this syndrome (i.e., hypogonadism, azoospermia, gynecomastia, elevated average height and increased plasma gonadotrophins [3]), a linguistic impairment can often be found [1, 2]. Accordingly, the intellectual profile of KS patients, though appearing within the normal range [4], reveals a discrepancy between Performance and Verbal IQ level with the latter being lower than the former [5].

Language difficulties include delayed onset of language, causing children with KS to reach the acquisition of the language development milestones at a slower rate [6]. In adults, there are evidence about the presence of disfluencies associated to sounds and syllable articulation; additionally, problems are reported in words retrieval and verbal fluency [7].

Language impairment appears often associated to reading, writing and reasoning disabilities [7]. These difficulties are sometimes so severe that patients meet the criteria for a diagnosis of learning disability (i.e. dyslexia, dysorthography etc.) [8]. As a consequence, the academic skills of individuals with KS (as well as patients with sexual chromosome aneuploidies, [9]) might be dramatically affected by the presence of a learning disorder [10].

To sum up, the bulk of the studies suggest that patients with KS show difficulties in tasks requiring the use of language competence. The areas under investigation appeared mainly to be: phonetic and phonological articulation, language comprehension and lexical access. Importantly, to our knowledge, it has never been tested the pragmatic competence of patients with KS.

The pragmatic area is concerned with the social and contextual appropriateness in the use of language. In the present study we aim at fulfilling this lack. In order to investigate pragmatic skills in KS we considered one of the most common pragmatic decisions that speakers make when producing a sentence: namely, choosing a referring expression to refer to an entity in a sentence (i.e. a noun, a pronoun or a null pronoun). For instance, a speaker might use expressions such as names (as "*cat*" in "*the cat saw the dog*"), pronouns (as "*it*" in "*it saw the dog*") or null pronouns ("*the cat saw the dog and Ø ran away*"). In the current study, we tested this production process in adolescents and young adults

*Address correspondence to this author at the Department of Psychology, University of Milano-Bicocca, U6 Building, Piazza dell'Ateneo Nuovo 1, 20126, Milano, Italy; Tel: +39 (0)2 64483805; Fax: +39 (0)2 64483706;
E-mail: mirta.vernice@unimib.it

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with KS, in order to understand how it may differ with respect to a group of age-matched controls.

As the study has been conducted in Italian, we first provide an overview about the pronominal system in such a language. In its pronominal inventory, Italian includes two pronominal forms: overt pronouns (e.g. *lui*, *lei*, *loro*; respectively he, she, they) and phonetically null pronouns [11]. Overt and null pronouns involve distinct syntactic and pragmatic features: referents that are readily inferable in the discourse context tend to be expressed by means of null pronouns, whereas referents less accessible are referred to using highly informative expressions, such as overt pronouns. Therefore in a sentence such as: "Il gatto vede il cane e Ø scappa via" (lit. "*the cat saw the dog and Ø ran away*"), the hearer might easily infer that the null pronoun refers to "the cat". In contrast, a sentence like: "Il gatto vede il cane e il gatto scappa via" (Lit.: "*the cat saw the dog and the cat ran away*"), though still grammatical, would be interpreted as if another cat was running away, while the first cat was still looking at the dog. Thus, producing an explicit referential expression (i.e. a full noun phrase "the cat" or a pronoun "lui") in such a case, would possibly result in a violation of the sentence meaning, preventing the hearer from fully understanding what was going on in the scene. Thus, the appropriate use of null referential expressions appears to be a crucial pragmatic competence for an effective communication.

Thus, as a general rule, one might consider that referential expression might be null when they refer to an entity already introduced in the linguistic context, whereas they have to be explicit and more informative (full pronouns or names) when they are used to introduce a new referent, that is unknown to the hearer's mind.

As a consequence, the choice of a referential expression is based on how accessible a referent is not only to the speaker's mind, but crucially to the hearer's mental model [12]. For this reason, it has been proposed that the choice of a referential expression like a null or overt pronoun involves considering not only the syntactic conditions that license a certain pronoun in a linguistic context, but also the pragmatic conditions that determine its appropriateness in discourse [13].

In the current study we tested the referential choices made by adolescents KS and an age-matched control group of typically developing adolescents in a narrative task, based on the procedure of [14]. Each participant viewed a Sylvester and Tweety cartoon (Canary Row [15]), and told the narrative to the experimenter. We analyzed each reference to the two main characters: Sylvester the cat and Tweety the bird. We further calculated the frequency with which each participant used null referential expressions (null pronouns) out of all the references to that character. In addition, in order to control for possible effects of reduced cognitive resources during utterance planning we further compared the Mean Length of Utterance between the KS participants and their controls.

We are confident that understanding the specific language ability underlying this referential choice process is important for the development of a language phenotype of KS.

METHOD

Participants

Eight KS patients participated in this study: 7 of them had a karyotype 47, XXY, whereas only 1 participant had a karyotype 48, XXYY. Their Mean age was 18; 25 (years; months), SD=2;6. All of them were under testosteron treatment. Three of them had received a diagnosis of Dyslexia. Eight control participants matched for chronological age (± 3 months) took part in this study [Mean age = 18; 37 (years; months), SD=2;36]. For each participant we tested the cognitive level, the receptive vocabulary and the comprehension skills by means of a set of cognitive and linguistic tests, namely the Raven Progressive Matrices, the Vocabulary task of the WAIS-R and the Token Test.

Procedure

The procedure followed basically that reported in [14]. We exposed participants to a Sylvester and Tweety cartoon, The "Canary Row", that has been used in literature to elicit narratives and gestures [15]. The cartoon involves a story in which Sylvester the cat tries to catch Tweety the bird, who is protected by its owner, Granny. The cartoon was proposed to the participants into three segments to reduce the memory load. Each segment lasted around 2 - 3 minutes. After each segment, participants were asked to retell the story to the experimenter. Their narratives were recorded and scored off-line.

Coding

Each narrative was divided into clauses, which could include only a main clause or a main and a subordinate clause. For each clause, a coder recorded

the number of words and the type of referential expressions used. A different coder excluded those cases that involved acoustically unclear or ungrammatical sentences. Referential expressions were coded by both coders as one of the following categories: null pronoun *("e Ø corre via")*, full pronoun ("*lui*, *lei"*), name (The Italian equivalent of Sylvester and Tweety, namely "*Silvestro* e *Titti"*). The proportion of explicit referential expressions was then contrasted against the proportion of null pronouns used appropriately. To do so, we first collapsed the amount of explicit referential expressions (i.e. full pronouns and names) into a single variable. Then, we created a categorical dependent variable, which contrasted proportions of null pronouns (expressed as 1) out of all other explicit referential expressions (i.e. full pronouns and names), expressed as 0. As our dependent variable is categorical, we submitted our data to a mixed effects logit model [16].

RESULTS

Cognitive Level

In this section, we report the results of the standardized tests. KS participants were administered a full Intelligence scale (i.e., WAIS). They showed a Total IQ = 90 (SD=8.89), a Verbal IQ= 92 (SD=9.22), and a Performance IQ = 88 (SD=10.45). Therefore, in our sample, we did not observe the discrepancy between Performance and Verbal IQ level, often reported in the literature [3].

We further tested KS and control groups on a range of cognitive and linguistic tests. The intelligence level was evaluated in both groups by means of the Progressive Matrices (PM) of Raven. The Token test was used as a measure to assess receptive language skills. The mental lexicon was investigated by means of the Vocabulary Task of the WAIS-R.

The progressive Matrices, the Token test and the Vocabulary Task raw scores for KS participants and controls were converted in z-scores (using the Italian standard norms for the relevant chronological age). Results are reported on Table **1**.

Participants with KS obtained overall lower z-scores than controls, although the difference was less evident in the non-linguistic test, namely, the PM of Raven. Additionally, the performance to the three tests varied to a different extent in the two groups: in the KS group the Vocabulary task elicited the lowest accuracy, followed by the Token test. In contrast, in the control group the Vocabulary task involved the highest accuracy level.

We performed a t test on the proportions of z scores in order to compare the results achieved in each test by KS participants and their age-matched controls. The analysis revealed that KS differed with respect to controls only in the scores on the Vocabulary Task [$t(7) = 4.77$, $p<.001$]. Scores at the Token test [$t(7) = 1.95$, $p=.09$] and at the PM Raven [$t(7) = 1.52$, $p=.17$] did not differ. A similar pattern of results suggests that the two groups did not differ in the non-linguistic task (PM of Raven), whereas their difference was more consistent in the linguistic tasks, with KS participants obtaining significantly lower scores in comparison with controls.

Referential production and Mean Length Utterance (MLU)

The utterances were transcribed and then analyzed. In the final dataset, for each participant we had a set of clauses, that constituted the dependent variable. For each clause we specified whether it involved a null referential expression (reported as 1) or an explicit referential expression (reported as 0). We submitted the data to a mixed effects logit model. As for the fixed factor (the independent variable) we included Group (KS vs. Control).

We observed that, on average, KS individuals produced less null referential expressions (43%; SD = 23%) than controls (48%; SD = 33%). However, the difference was only marginally significant (Wald $Z=1.81$, $p=.07$). That is, KS individuals did not significantly differ from their age-matched controls in the ability to appropriately produce null referential expressions.

Table 1: We Report Means and Standard Deviations (in brackets) of the z-Scores in the Three Standardized Tests by Participants with KS and Controls

	Token Test	PM Raven	Vocabulary Task
KS	-.38 (1.35)	.33 (.43)	-.91 (1.26)
Controls	.61 (.74)	.81 (.85)	1 (.68)

Furthermore, for each participant, we calculated the length of the sentences (MLU), expressed in count of words. As we are dealing here with a "count of" variable, we performed a Poisson regression [17] including count of words per clause (MLU) as dependent variable, and group as independent variable. The utterances of KS participants were found to be shorter than that of controls (KS: 6.10 words ± 2.84; controls: 7.90 ± 1.41), but again, also this difference did not result significant (Wald Z = - .70, p >.05).

Finally, regression analyses were carried out to study correlations between MLU and scores on linguistic (Token test and Vocabulary task) and cognitive (PM Raven) tests. Interestingly, the score at none of these tests appeared to predict MLU (all ps >. 28). We could thus conclude that MLU was not modulated by the general cognitive and linguistic level of the speakers.

DISCUSSION

In the current study we aimed at evaluating the pragmatic competence of patients with KS, by comparing them with a group of chronological age-matched controls. We did so by testing the ability of individuals with KS to appropriately produce referential expressions in a narrative.

The pattern of findings indicates that the reference form choices of participants with KS and typically developing controls were comparable. Descriptively, KS patients appeared to be more likely to use explicit expressions than controls, however the difference was only marginally significant. Therefore, our results support the idea that KS were sensitive to discourse and pragmatic constraints almost to the same extent as controls.

Also the MLU appeared to be comparable for KS and control groups. Indeed, even though sentences in the KS group appeared to be on average shorter as compared to those in the control group, the difference was not significant. One possible explanation for the lack of effect found in this variable could be that MLU might provide useful information about language development only in children, but that it is an inadequate measure of linguistic complexity above a certain age.

We believe that the most prominent difference between groups, emerged in the performance to the standardized tests: participants with KS were significantly below their age-matched controls in the Vocabulary task, a measure of their lexical abilities, and marginally below controls in the accuracy to the Token Test, a task that evaluates receptive skills. In contrast, the non-verbal reasoning abilities, tested through the PM of Raven, stand out as an area relatively preserved for participants with KS. Such pattern of results confirms again that linguistic tests are particularly taxing for this population. This is not surprising, as in the literature it is reported a well known discrepancy between verbal and non-verbal abilities [2-4].

Moving from the assumption that the two groups did show a difference in their linguistic performance, at least according to their accuracy in the standardized tests, one could ask why we did not see greater differences between the two groups in the production of referential expressions.

One possible answer could be that reference production is more than a purely linguistic task, involving higher-level cognitive abilities, beyond language. Indeed, choosing an appropriate referential expression requires the activation of a number of cognitive cues, among which attention and memory [18]. Therefore, one could conclude that the choice between null pronouns and more explicit expressions involves more complex processes than one could suppose.

In conclusion, although our participants with KS showed some linguistic deficit, as attested by their performance to the standardized tests, they appeared to master the pragmatic competence required for an adequate reference production. That is, individuals with KS appeared to be able to communicate effectively by using appropriate referential expressions.

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