

## Cognitive Aspects of Fetal Alcohol Syndrome in Young Adults: Two Case Studies

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### ABSTRACT

Fetal Alcohol Syndrome (FAS) includes a specific pattern of cognitive deficits, including language and speech disorders. Two adult participants were studied by means of the Examination of Language Test and a battery of complementary tests to evaluate oral and written language. Our data indicated great results of the participant who had psychopedagogical and speech-language assistance than the other who did not receive none intervention. These findings suggest that early diagnosis and intervention are crucial to minimize sequelae in FAS.

*Keywords:* sexuality; fetal alcohol syndrome; language evaluation; early intervention.

### RESUMO

#### Aspectos Cognitivos da Síndrome Fetal Alcoólica em Adultos Jovens: Dois Estudos de Caso

A Síndrome Fetal Alcoólica (SFA) inclui um padrão de déficit cognitivo, incluindo alterações de linguagem e fala. Estudou-se dois participantes adultos que foram diagnosticados com SFA na infância por meio do Teste de Exame de Linguagem e de uma bateria de testes complementares para avaliar aspectos da linguagem oral e escrita. Os dados indicaram que o participante que recebeu uma intervenção psicopedagógica apresentou melhores resultados que aquele que não tinha recebido nenhuma intervenção. Estes achados indicam que um diagnóstico precoce é fundamental para minimizar as sequelas da SFA.

*Palavras-chave:* Síndrome fetal alcoólica; avaliação da linguagem; intervenção precoce.

Alcohol is the most widely used psychotropic drug worldwide. Approximately two thirds of the population of Western countries consumes alcoholic drinks at levels above occasional use (WHO, 2004) and alcohol dependence ranks 5<sup>th</sup> among the main health problems for individuals aged 15 to 44 years worldwide (WHO, 2009). In Brazil, the prevalence of alcohol dependence is high, varying between 17.1% in males and 5.7% in females (Galduroz, Noto, Nappo, & Carlini, 2005). Among the various problems associated with the abuse of alcohol, its consumption by pregnant women may lead to Fetal Alcohol Syndrome (FAS) (Jones, 2011). The initials descriptions of this syndrome were provided by Lemoine, Harousseau, Borteyru and Menuet (1968) as well as Jones & Smith (1973) and includes clinical findings, such as growth retardation before and after birth, dysfunction of the

central nervous system and abnormalities of the face. Lemoine et al. (1968) described 127 children with specific facial features from prenatal alcohol exposure and other symptoms described above. Other malformations affecting the eyes, including low visual acuity, were later reported (Stromland & Sundelin, 1996).

Alcohol has teratogenic properties and causes congenital malformations in fetuses (Godin et al., 2010). Moreover, alcohol alters the function of the placenta by causing contraction of the blood vessels and decreasing the exchange of oxygen and nutrients between the mother and the fetus (Perez, Velasco, Monte, Gonzalez-Buitrago, & Marin, 2006). Teratogenic substances affect the fetus in different ways depending on the developmental stage. Some substances used at the beginning of pregnancy (up to 16

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days after fertilization) may have an all-or-nothing effect, *i.e.*, they either result in the termination of pregnancy, or they do not affect the fetus at all. At this stage, the fetus is highly resistant to congenital defects. However, between the 17<sup>th</sup> and 57<sup>th</sup> day after fertilization, the fetus is particularly vulnerable to such defects because its organs are developing at this time (Gemma, Vichi, & Testai, 2007). According to some authors, teratogenic effects appear only when alcohol is consumed during the first trimester of pregnancy. Nevertheless, the development of the central nervous system remains vulnerable and is at a higher risk between the 12<sup>th</sup> and 18<sup>th</sup> weeks (Gemma et al., 2007). Generally, alcohol impairs the correct migration of neurons to specific brain areas leading to serious cognitive impairments because this migration depends from glial cells. Because these cells would be unprovided of nutrients (alcohol leads to anorexigen effects), they could not help the neurons to reach the correct areas (Kumada, Jiang, Cameron, & Komuro, 2007). Hence, alcoholic women risk giving birth to children with FAS.

The prevalence of FAS is about 2.8-4.6/1,000 births in the USA, and it rises to 9.1/1,000 when the cases of children with neurological developmental disorders are considered a spectrum of FAS. In Italy, the prevalence of FAS is 3.7-7.4/1,000 births (May et al., 2006). However, in developing countries, such as South Africa, the numbers are very high, and the rate varies between 68 and 89.2/1,000 births (May et al., 2007). In Brazil, Mesquita and Segre (2009) evaluated 1964 newborns in the city of São Paulo and the prevalence of FAS symptoms was 38.7/1.000 births. Interesting, the same authors detected that three months before the pregnancy around 40% of women consumed alcohol weekly and during the gestation this number was near 20% and decreased in the first, second and third semester respectively (21.2%, 17.5% and 17.1%). Momino, Felix, Abeche, Zandona, Scheibler, Chambers, et al. (2012) compared two sample of adolescents and/or young adults (institutionalized male adolescents *versus* male students) in the Porto Alegre city and observed that almost 49% of the mothers of institutionalized adolescents admitted to drink during the pregnancy compared with 40% of the second group. In addition, the same authors observed that FAS symptoms were more common in the institutionalized male group. Importantly, these numbers vary not only as a function of the socio-economic conditions in each country but also according to the diagnostic methods employed (May et al., 2009). The

exact amount of alcohol consumed by pregnant women is difficult to determine, and many young women are likely to report less than they actually consume (Monino et al., 2012).

There are a number of data showing that many children with FAS have problems in language acquisition and others language impairments (Church & Kaltenbach, 1997). However, there are few studies evaluating specific language impairments in children with FAS and some of those studies were conducted using native American or African-American children, restricting the data observed (Manji, Pei, Loomes, & Rasmussen, 2009). Children with and without FAS differ with respect to the grammatical and semantic aspects of language and memory skills (Church & Kaltenbach, 1997). In general, children with FAS have limited morphological or syntactic understanding, and they have difficulty understanding verbal ditto. In addition, they show limited understanding of isolated words belonging to domains they already know because the memory skills, are less able to retain linguistic elements through short-term memory (Becker, Warr-Leeper, & Leeper, 1990). Some authors have observed that children with FAS exhibit a significant delay in the acquisition of language as well as in receptive and expressive language in addition to impairment in their syntactic, grammatical and semantic abilities (De Beer, Kritzing, & Zsilavec, 2010), and they are at a higher risk for hearing loss (Church & Kaltenbach, 1997).

Although many children with FAS show language and cognitive impairment, early interventions are associated with decrease risk of adverse outcomes (Peadon, Rhys-Jones, Bower, & Elliott, 2009). There are a wide range of interventions available in children with FAS, such education and learning strategies (Coles et al., 2007), speech-language therapy (Timler, Olswang, & Coggins, 2005) and training of social skills (Rasmussen, Becker, McLennan, Urchuk, & Andrew, 2011). For the reasons cited above, the aim of this study was to evaluate oral and written language skills in young adults with FAS.

## METHOD

Participants in this study were two adult male individuals who were 17 and 21 years of age and residents in the city of Sao Paulo. Both were diagnosed with FAS in early childhood at the Department of Genetics at Universidade Federal de São Paulo-UNIFESP. They were diagnosed by following the guideline from

National Research Council (1996) based on the nuclear symptoms such: growth retardation, neurodevelopment impairment and facial dimorphism (at least two characteristics must be present such microphthalmia or short palpebral fissures, maxillary hypoplasia, microcephaly or thin upper lip), intelligence and behavior impairments. During childhood and adolescence, one of them received psychological and speech-language assistance and attended a regular school; the other subject had no assistance whatsoever and attended school only up to the 4<sup>th</sup> grade of elementary school.

## Procedures

After initial contact with the Department of Genetics at the Universidade Federal de Sao Paulo, we surveyed its database of patients diagnosed with FAS since 1980. Ten patients were identified among the clinical records, and we sought to schedule interviews with them. However, only two patients had remained at the same address and had the same phone number as stated in the records, we could not locate and interview the other eight patients. Initial contact with their caretakers was conducted by telephone. The researchers identified themselves, offered a description of the study and invited each participant to participate by scheduling an interview. The interview sessions lasted 90 minutes and included the patient, his caretaker and two speech-language researchers. Each subject signed an Informed Consent from during the first interview (approved by the Committee for Research Ethics of UNIFESP – CEP 0756/006) and was then invited to participate in the first stage of the speech-language evaluation to assess oral and written language. A second interview was carried out one week later to complete the speech-language evaluation and to communicate the results to the participants and caretakers.

## Instruments

The following instruments were used for the speech-language evaluation:

1. The Examination of Language Test TIPITI (Braz & Pellicciotti, 1988). The Examination of Language Test TIPITI includes the following areas: Categorization of words: participants chose one word among four that was not semantically related to the others and were asked to justify their choice. Participants had to recognize traits and attributes relevant to the recognition and organization of semantic classes. Definition of words: participants were asked to define several words. Interpretation of the results sought to

assess the manner by which subjects analyze language, which attributes they consider relevant, how they order such attributes, and on what level of generalization and specialization they operate. Immediate auditory memory: participants were asked to listen to a sentence or a sequence of words and then to repeat it. The aim of this activity is to assess short-term memory. It is expected that adolescents who are 15-18 years old will be able to memorize five disyllabic, trisyllabic and polysyllabic words belonging to the same semantic category and sentences with at least 28 syllables. Completion of sentences: participants had to finish a sentence spoken by the evaluator in a way that was syntactically and semantically appropriate. Interpretation of the results sought to evaluate performance at the morpho-syntactic and semantic levels. Reading of words: participants were asked to read regular, irregular and pseudo-words. It is expected that adolescents will be able to read all required words without any failure in decoding graphic symbols.

Reading of text: participants were given a text to read aloud. From the 3<sup>rd</sup> grade onwards, children are typically able to read through texts of significant length with prosody that is compatible with the punctuation.

2. The routine evaluation protocol regarding aspects of language applied at the Center for Assistance and Support of Adolescents, including the following tests: dictation: participants were asked to write regular, irregular and pseudo-words. It is expected that from the 3<sup>rd</sup> grade of elementary school onward, alterations such omissions of graphemes will decrease and become rare in adolescence. Free diction: participants were asked to write a short paragraph on the subject of their choice on a sheet of paper. Interpretation of speech is grounded on the principle of the existence of an interlocutor to whom the message is addressed. Oral understanding: participants were evaluated throughout the period of testing based on their responses to questions that were asked and their understanding of the explanations of the suggested activities. Written understanding: after reading a text in silence, participants were asked to retell the story. If the participant presented difficulties, questions were asked about the story that was read. Interpretation of the results sought to assess the understanding of written materials, and it took into account the level of schooling of the participants. Phonological awareness: participants were instructed to perform syllabic and phonemic segmentation, rhyme and perform phonemic synthesis, exclusion and transposition. It is ex-

pected that participants will correctly complete at least four or five of each of these items.

## RESULTS AND DISCUSSION

*Case I:* ECO, a 17-year-old male at the time of the interview, attended the 8<sup>th</sup> grade of elementary school. According to his clinical records, his mother was intoxicated most of the time and entered an alcoholic coma when she was 7 months pregnant. Among his physical traits were palpebral ptosis, bilateral convergent strabismus, small chin and a small labial philtrum. Because his neuro-psychomotor development, he started walking when he was three years old and was toilet trained at age five. He was diagnosed with mild bilateral sensorineural hearing loss. During the interview, his caretaker stated that ECO had frequent seizures and epilepsy all throughout childhood and adolescence. He received psychiatric treatment and used valproic acid and phenytoin.

*Speech-language evaluation:* In the word categorization task, he correctly categorized ¼ word for the 11- to 14-year-old age range and only 1/4 for the age range above 15 years old. Despite his two correct responses, he was unable to justify his choice. In the definition test, he was able to define all words, but his definitions were poor and based on personal experience. In the phonological awareness task, he correctly performed syllabic synthesis, phonemic synthesis and rhyming. For the phonemic exclusion and phonemic transposition tests, the subject was not able to segment any of the required words into phonemes. In the sentence completion test, he completed 7/10 sentences with correct syntax in the 11- to 14-year old age range and did not understand the conjunction “however”, which is expected for that age range. In complementary tests appropriate for his age (above 15), the subject did not understand any of the conjunctions used. In the immediate memory test, he could not repeat sentences with 26 syllables and only repeated one sentence with 22 syllables. Regarding graphic emission, omission and addition of letters were observed as well as incorrect use of graphic accents and mistakes in multiple representation and auditory exchanges. ECO was not able to complete the free writing task. When reading words and text, his reading speed was slow, syllabized and characterized by morphological permutations, omissions, additions, contamination and letter permutations. During the sessions, ECO understood all commands and suggested activities. However, during spontaneous conversation, he often had difficulty maintaining attention to the subject and

exhibited alterations in oral understanding. When assessing written understanding, he was not able to retell any part of the written text or correctly answer any of the questions about it.

*Case II:* PLGN, a 20-year-old male, attended school up to the 4<sup>th</sup> grade of elementary school. At the time of the interview, he worked in recycling collection. His clinical records reported that his mother smoked 20 cigarettes/day and drank one bottle of distilled alcohol/day until the 7<sup>th</sup> month of pregnancy. PLGN had meconium aspiration. He began speaking words when he was 1 year, 2 months of age and walking when he was 1 year, 6 months of age. Among his physical traits, he presented palpebral ptosis, malformation of the ears, a thin upper lip and a low nasal bridge.

*Speech-language evaluation:* in the categorization evaluation, PLGN did not understand the suggested activity, even when it was explained and exemplified. In the definition test, he was not able to define the noun “mountain”. When asked to define the other nouns, he was able to describe the use of the object using only two words, and he only mentioned attributes of the remaining nouns. His definitions were predominantly based on his personal experience. On the sentence completion test for the 5- to 7-year old age range, he showed difficulties using the conjunctions “until”, “but”, “therefore”, “however”, “rather than” and “unless”. In some sentences there were syntactic alterations. In the phonologic awareness task, PLGN correctly performed syllabic synthesis, one phonemic synthesis and one phonemic exclusion. He performed no phonemic transpositions, he could not segment any of the required words into phonemes and was not able to correctly identify any rhymes. For the working memory test, he did not repeat any sentences with 10 syllables. He did not repeat three polysyllabic words belonging to the same class, and he only repeated two words.

Regarding graphic emission, the following orthographic alterations were observed: incorrect use of the graphic accents, overgeneralization of rules and multiple representations. PLGN wrote a sentence instead of a paragraph. Alterations were observed in verbal concordance and incorrect use of graphic accents. Moreover, there was semantic alteration, as he showed a poor vocabulary and low linguistic mastery with respect to the elaboration of the text. For the task involving reading of words, he showed permutations of words. His reading of the text was mostly fluent. His reading appropriately reflected the punctuation,

but he read with little intonation, and at times his reading was syllabized. During the sessions, PLGN had difficulty understanding the suggested activities, and he did not coherently answer the questions posed by the evaluators. Regarding his written understanding, when asked to retell a text that he had read, he merely repeated one of the sentences and could not answer any of the questions about it, showing that he had not understood it.

This study sought to evaluate the effects of the use of alcohol during pregnancy and its implications for the development of oral and written language in patients with FAS. Both participants showed significant alterations in all aspects of their use and understanding of oral and written language that were evaluated in this study, thus confirming that the use of alcohol by pregnant women may cause irreversible damage to their children's intellectual development. The results of this study corroborate data reported by other authors showing that several developmental domains are susceptible to neurotoxicity from alcohol during pregnancy, including intelligence, language, reading, memory and attention (Autti-Ramo et al., 2006).

In our study, cognitive deficits of language were observed, including failure at tasks involving categorization, definition and immediate memory, thus indicating that language development is incomplete in these individuals. Our results are consistent with published literature, indicating that delays in the acquisition of language and deficits in receptive and expressive language might occur in individuals with FAS.

Garcia, Rossi and Giacheti (2007) observed in two brothers (one with 8 and other with 16 years old) the same characteristics such a verbal/nonverbal impairment comprehension in the city of Marilia (during the both pregnancies, the woman drunk daily around two distilled bottle). Both children lived under the same environmental conditions but the authors observed more intense symptoms in the older brother probably liked with higher levels of exposure during the specifics periods of pregnancy. Alterations in oral understanding, graphic emission and understanding, as well as the completion of sentences, indicate a failure in the communication system of these individuals. Moreover, alterations in phonological awareness indicate a lack of access to the phonological level of speech and the manipulation of phonemes, which are important stages in the learning of reading and writing (Bowey, Cain, & Ryan, 1992). Additionally, pragmatic alterations were observed, *i.e.*, a difficulty beginning or establishing a conversation, mastering

words and observing grammatical rules. The alterations that we observed suggest a speech-language diagnosis of delayed language acquisition and developmental disorder in both patients; the subjects did not acquire certain skills at the appropriate stages according to the normal process of acquisition of language, and the impairment then persisted throughout their development. Moreover, poor vocabulary and sentence structure were observed during conversation, suggesting an infantile type of behavior typical of younger age groups. Additionally, a learning delay was observed, as both subjects exhibited significant difficulty in keeping pace with their schoolmates.

Despite presenting similar alterations, case I (ECO) showed better results than Case II (PLNG). He was attending the 8<sup>th</sup> grade of elementary school at the time of the interviews, and he had undergone speech-language therapy since the age of 12. PLGN stopped attending school in 4<sup>th</sup> grade and never received any speech-language intervention. The comparison of the two cases suggests a significant influence of schooling and the importance of speech-language intervention for prognosis. Nevertheless, it must be emphasized that other factors might also influence the development of language, such as a suitable environment and stimulation by the family, in addition to the course of the syndrome, which varies among affected individuals (Peadon et al., 2009).

Participation of the family and an environment adapted to FAS patients are important to improving prognosis. Early diagnosis and intervention might help these individuals develop in a more typical manner by addressing their developmental deficits. In the speech-language field, early therapeutic intervention might contribute to improvements in the oral and written communication of patients, thus minimizing the sequelae arising from alterations in communication (Timler et al., 2005).

## CONCLUSIONS

Psychopedagogical and speech-language assistance seem to have been effective in the participant who received early intervention. These findings suggest that early diagnosis and intervention are crucial to minimize sequelae in FAS. Furthermore, health-care professionals must pay special attention to the use of alcohol by pregnant women, informing them about the effects of alcohol on fetal development, in addition to answering further questions.

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