AROUSAL DIFFICULTIES IN MALES WITH FRAGILE X SYNDROME

By Vicki Sudhalter, PhD and Richard Belser, PhD
New York State Institute for Basic Research in Developmental Disabilities
1050 Forest Hill Road
Staten Island, New York 10314

Introduction

It has been reported that males with fragile X syndrome [fra(x)] have characteristic physical features and are especially prone to certain health problems, such as certain types of seizures, and otitis media. They may also exhibit schizoid or schizotypal behaviors more often than their peers with similar behavioral and cognitive challenges not caused by fra(x). Despite these reported differences, a long standing controversy has existed as to the importance of the fra(x) diagnosis when considering the behavioral and cognitive disabilities of affected individuals. For example, some professionals in the field believe that individuals with fra(x) who exhibit autistic or attention deficit behaviors are indistinguishable from those with similar behavioral disorders who do not have fra(x).

We strongly disagree with this position, and believe that individuals with fra(x) do exhibit distinctive phenotypic features that differentiate them from other individuals. Specifically, we believe that the single most important trait of the fra(x) syndrome is a difficulty in modulating arousal, and that this inability to normally modulate arousal is directly responsible for many of the distinctive behaviors found in fra(x).

Although there has been speculation within the clinical community that the abnormal response to sensory stimulation seen in fra(x) has some relation to anxiety or arousal, very little research has been done to investigate such a role of arousal in fra(x). It is our belief that the inability to normally regulate arousal provides a useful framework for understanding many of the behavioral symptoms of fra(x), and we are beginning to acquire experimental evidence that such a dysfunction does exist in this population.

It is clear from observing the daily behavior of people with fra(x) that they tend to be bothered by a wide variety of arousing stimuli, including loud sounds, bright or flashing lights, visually complex or crowded environments, unpredictable events and the maintenance of eye contact. Those with fra(x) typically respond to such stimuli by making avoidant behaviors, which include covering their eyes or ears, and exhibiting tactile defensiveness and gaze aversion. When appropriate means for coping with this arousal are not available, it may lead to self-abusive or aggressive behavior.

Problems with anxiety disorders, especially social anxiety, have been well documented within the fra(x) population, as has aversion to eye contact. Social avoidance, which refers to a set of related behaviors that includes gaze aversion, tactile defensiveness, overt turning away of the body during face to face social interaction, and stylized, highly ritualistic forms of greeting, has been shown to be evident in individuals with fra(x). In fact, the avoidance of social contact appears to distinguish fra(x) males with autism or atypical pervasive developmental disorder (PDD) from non-fra(x) males with similar developmental disabilities. For example, fra(x) males with PDD and autism have been found to be more avoidant with strangers than non-fra(x) autistic or PDD males. Fra(x) males, with and without autism, also have been shown to be more likely than non-fra(x) MR males, with and without autism, to selectively avoid eye contact and not establish mutual gaze.

In order to provide empirical evidence to support the clinical observations of arousal dysfunction, we are currently studying the relationship between arousal and both verbal and non-verbal behavior in fra(x). The following sections summarize some of our recent findings, and discuss their potential implications for education.

Non-verbal measures of arousal

We are currently studying anxiety in individuals with fra(x), as well as their non-fra(x) peers with autism, developmental delay and attention deficit disorder. Because there are very few ways to assess anxiety within a population with developmental delays, clinicians must infer what these individuals are experiencing on the basis of their overt behavior. Unfortunately, such inferences can be confused by the behaviors normally associated with such diagnoses as autism, PDD or attention deficit hyperactivity disorder (ADHD), as well as by IQ level. For example, some of the nonverbal behaviors emitted by individuals with fra(x), such as perseveration, avoidance of eye contact and difficulty with transitions, could easily lead to misdiagnoses by clinicians who do not fully appreciate the effects of arousal and anxiety in this population. Thus, in order to most appropriately treat these individuals by recommending effective intervention programs or medicating the correct symptoms, it is important to be able to distinguish behaviors indicative of anxiety disorder from behaviors which reflect other disorders.

Our studies have consistently found that males with fra(x) exhibit significantly more anxious behavior during conversation than do non-fra(x) cohorts matched for age and adaptive level. This indicates that the anxious behavior seen in males with fra(x) is not solely a consequence of age or developmental delay. In one study, the social anxiety behavior found to be most closely associated with fra(x) was the lack of appropriate communicative gestures. This is consistent with previously reported work by other researchers which showed that such a lack of communicative gestures was able to discriminate males with fra(x) from others.

A more recent study from our laboratory focused on social disengagement, the tendency to actively avoid eye contact by turning away from another person during one-on-one conversation. We found that social disengagement clearly distinguishes males with fra(x) from cohort samples composed of individuals with MR, autism and attention deficit disorder not associated with fra(x). We believe that this social disengagement is one way that individuals with fra(x) cope with the uncomfortably high anxiety that they experience during close interpersonal interactions.

These recent studies have begun to experimentally establish the fact that anxiety does distinguish males with fra(x) from others with developmental delay, autism and attention deficit disorder. In order to obtain additional evidence we are continuing this research with more individuals, and are refining our behavioral measurement scales.

Verbal measures of arousal

In addition to non-verbal behaviors, speech and language problems have also been described in many studies of males with fra(x). Deviant verbal behaviors include tangential language, exemplified by the speaker's inability to maintain topic, and perseverative language, exemplified by repetitions of words, phrases and topics. The presence of perseverative language is one of the most characteristic language deviances produced by males with fra(x), and is the verbal behavior that best discriminates them from non-fra(x) males with autism and MR.

Research currently being performed by ourselves and our colleagues at the Institute for Basic Research, is revealing an association between eye gaze and the probability of deviant language production among males with fra(x). During interactions with both familiar and unfamiliar adults, we found a significant difference in the probability that males with fra(x) would produce deviant repetitive language when adults looked at them while they talked, compared to when the adults turned away. These results were interpreted as suggesting that heightened sensitivity to social gaze caused the males with fra(x) to feel discomfort, which then led to an increased production of atypical repetitive language. We believe that the "discomfort" thought to underlie the link between

eye contact and language deficits is essentially anxiety, which directly reflects heightened arousal.

Discussion

We have summarized the results of several preliminary studies that suggest that males with fra(x) exhibit significantly more anxious behavior during conversation than do their non-fra(x) peers, and are more physiologically aroused by eye contact with their conversational partners than are non-fra(x) males. Thus, it appears that hyperarousal, and hyperarousability, are related to fra(x) independently of developmental delay.

Although more studies are required in order to generalize about the possible role of arousal dysfunction in the etiology of fra(x) behaviors, our results do support the hypothesis that the maintenance of eye contact during conversation can be a significant source of arousal for males with fra(x), which may in turn underlie the production of social avoidance and deviant language.

This hypothesis has clear implications for improving social interactions with individuals who have fra(x). For example, it may be more appropriate to react to their perseverative and dysfluent speech by attempting to identify and deal with any sources of underlying anxiety, rather than simply modeling correct language production, since such pressure could easily exacerbate their problem.

In addition, our results and theoretical model have very important implications for the assessment and treatment of anyone with fra(x), and should be taken into consideration when interpreting their behavior and drawing conclusions about their cognitive and linguistic competence. Performance on standardized tests, for example, may be depressed due to overarousal and anxiety caused by the test situation. To assure the most accurate assessment of their aptitudes, these individuals should be made as comfortable and relaxed as possible during any testing. They should be allowed to initiate eye contact, rather that having it forced on them during conversation or speech therapy, and greater attention should be paid to reducing potentially stimulating aspects of their environment.

Our model also suggests that intervention strategies based on relaxation and anxiety reduction techniques may be especially effective for those with fra(x). By teaching these individuals to more effectively cope with stress, it may be possible to reduce their anxiety, and their production of deviant language, thus promoting a more comfortable affect, better communication and an improved quality of life.