

The Impact of FXTAS on Patients and Their Spouses

Receiving a diagnosis of fragile X-associated tremor/ataxia syndrome (FXTAS) can be overwhelming for everyone in a family unit. Patients, their spouses and family members often have many questions about the diagnosis, clinical features, treatment, interventions, and planning. Many of these questions can be answered by a neurologist and primary care physician, but other providers are often needed to address a wide range of concerns.



Liane Abrams

Given the limited experience many physicians have with this newly described disorder, they may be unable to address issues outside their area of expertise, particularly those involving long-term care, prognosis, and planning for the future. If your family member has FXTAS, it will be very helpful if you have access to one of the Fragile X clinics around the country (see www.FragileX.org/clinics). It can also help to provide your physician with publications that might enhance his or her knowledge of FXTAS (see resources listing at end of this article).



Louise Gane

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Anxiety and the emotions that accompany the diagnosis can contribute to this difficulty. FXTAS can include cognitive and emotional impairment in the patient, which can also interfere with communication at the time of diagnosis.

The ramifications of this diagnosis affect the patient and spouse differently, with different fears and concerns. It is common for patients to react to the diagnosis with denial, anger, grief, loss of self-esteem, and fear of the future. They may feel guilty or sad about their physical changes and they may lack the means to express feelings of loss of control, lowered self-esteem, and being deprived of an enjoyable retirement.

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Spouses may experience depression, isolation from friends and family, grief, anger and frustration over the possible loss of the future the couple planned together. It is typically the spouse who must anticipate the future needs of the patient. Because spouses usually fill the caregiver role, it is important for them to take care of themselves, both physically and emotionally.

Sometimes the medical information and all its ramifications are overwhelming. It can be very difficult for the individual and spouse to hear that the patient may have to stop driving, use a walker, or will become incontinent. The loss of independence and the change in expectations for the future can be shattering.

It is particularly difficult when the genetic aspects of the FMR1 premutation have to be addressed with children, grandchildren or other family members. If there are grandchildren with fragile X syndrome, it can add emotional stress to the family to realize there is another family member affected. It may be helpful to consult a genetic counselor who is familiar with the issues of inheritance and carrier testing for family members, as well as the stresses of having more than one affected family member with multigenerational effects of the FMR1 gene.

It may also be helpful for the patient and spouse to meet with a social worker or family therapist to process these emotions and gather support and information to deal with the realities of the situation.

Sometimes the patient and spouse have different perceptions of how FXTAS has affected the patient. The spouse may find the patient irritable, moody, disinhibited, or showing other personality changes. The patient may feel these changes are in response to the diagnosis, not caused by the disorder. It is common to feel angry or frustrated following a


FXTAS diagnosis, yet personality changes can also be part of the condition. It may be difficult to discern which behaviors are related to the clinical presentation of FXTAS and which are a response to the diagnosis.

For the spouse, creating a network of family, friends and activities outside the home can help create independence and an identity beyond that of “caregiver.” It can be helpful to establish routines such as attending community or religious functions, playing bridge, engaging in regular exercise, and maintaining relationships that involve time away from caregiving duties. This also creates a network of support should the spouse find him- or herself alone. These routines help caregivers maintain emotional health and become more effective in their role. Though they may feel guilty about leaving their spouse, the rewards for patient and caregiver outweigh any negatives.

The spousal caregiver may need physical help with the everyday needs of the FXTAS patient, such as weight-bearing, lifting, cleaning, feeding, dressing and bathing. Many families may find themselves relocating to a single-level house or apartment, purchasing a van and putting in a lift, installing bars in the bathroom, and hiring health care aides.

The caregiver may need additional support and counseling if the FXTAS patient is experiencing increased irritability or mood changes.

If symptoms of FXTAS progress, the long-term needs of the patient must be considered. Financial planning, insurance coverage, health care directives, and end-of-life care should be discussed with appropriate professionals and other family members. Local agencies that serve patients and families who have chronic or degenerative conditions should be contacted; they can help with important health care and social services and referrals to other resources.

In addition, The National Fragile X Foundation is here to help. We understand how important it is to have resources, support, guidance and education to deal with this complex disorder. Feel free to call at 800-688-8765 for more information, or consult the resources below. 

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RESOURCES For additional information please visit the FXTAS section at www.FragileX.org and look for the following articles listed near the bottom of the page:

- Fragile X–Associated Tremor/Ataxia Syndrome: An Aging Face of the Fragile X Gene (2008)
- Treatment of Fragile X-associated Tremor/Ataxia Syndrome (FXTAS) and Related Neurological Problems (2008)
- The Five Most Common Questions About FXTAS (2007)
- FXTAS Q&A
- Fragile X-Associated Tremor/Ataxia Syndrome (FXTAS)
- Understanding FXTAS: Causes, Symptoms, Diagnosis, Research
- The ABCs—and Xs—of FXTAS Genetics
- Fragile X’s Unwelcome Relative
- The Identification of FXTAS in Rare Carrier Females
- Fragile X Premutation Tremor/Ataxia Syndrome: Molecular, Clinical, and Neuroimaging Correlates (2003)
- Fragile X-Associated Tremor/Ataxia Syndrome: Clinical Features, Genetics, and Testing Guidelines
- FXTAS: A Progressive Neurologic Syndrome Associated with Fragile X Premutation (2005)
- Fragile X Premutation Tremor/Ataxia Syndrome: Molecular, Clinical, and Neuroimaging Correlates (2003)
- Bacalman, S., Farzin, F., Bourgeois, J. A., et al. (2006) Psychiatric phenotype of the fragile x-associated tremor/ataxia syndrome (FXTAS) in males: Newly described fronto-subcortical dementia. *J Clin Psychiatry* 67(1): 87-94.
- Gane, L.W., Flynn, L. and Hagerman, R.J. (2005) Needs and priorities of fxtas patients and spouses determined using q-sort methodology. The 12th International Workshop on Fragile X and X-Linked Mental Retardation, August 26-29, 2005, Williamsburg, VA.

FXTAS Listserv:

To join, go to <http://health.groups.yahoo.com/group/FXTAS/> and select the “Join” option.