FRAGILE X GENE MUTATIONS AND PREMUTATIONS:
PSYCHIATRIC MANIFESTATIONS

Aims and Scope:
Full mutations of the fragile X mental retardation \textit{FMR1} gene (over 200 CGG repeats) lead to the fragile X syndrome (FXS), usually recognized during childhood. FXS may be associated with mental retardation, autism spectrum disorders, and varied behavioral disturbances. Less well known, \textit{FMR1} gene premutations (55-200 CGG repeats) may manifest clinically as primary ovarian insufficiency in up to 20\% of women carriers, or as fragile X-associated tremor/ataxia syndrome (FXTAS) in both genders. A host of related medical, endocrine, and immunological problems are common in carriers of the \textit{FMR1} gene premutations as well. FXTAS is more prevalent in men, although women may be affected. FXTAS is associated with a high lifetime prevalence of depressive disorders (65\%) and anxiety disorders (52\%). Cognitive disturbances in premutation carriers include executive dysfunction and deficits in short term memory, attention, working memory, declarative and procedural learning, and capacity for response inhibition. In later stages of FXTAS, dementia may occur in up to 40\% of men, causing a serious caregiver burden. In this issue, we will review the psychiatric manifestations of FXS and FXTAS and implications on affected families.

Key words:
Fragile X syndrome (FXS), fragile X-associated tremor/ataxia syndrome (FXTAS), \textit{FMR1} premutation, psychiatric, genetics

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Schedule:
- Manuscript submission deadline: March 1, 2012
- Peer Review Due: April 1, 2012
- Revision Due: May 1, 2012
- Notification of acceptance by the Guest Editor: June 1, 2012
- Final manuscripts due: June 15, 2012