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Fragile X Syndrome Genetics

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Message from the Guest Editors

Fragile X syndrome (FXS) is the most common inherited form of intellectual disability and the leading single-gene cause of autism. FXS is usually caused by a CGG trinucleotide expansion to greater than 200 repeats, inducing epigenetic silencing of the fragile X gene, FMR1, and loss of its protein product, FMRP, essential for normal neurodevelopment. Since the discovery of the causative mechanism of FXS around 1991, there has been rapid progress in better understanding FXS neurogenetics and developing improved diagnostic and screening techniques, as well as potential targeted therapies. Preclinical trials that assessed treatments for pathways dysregulated due to loss of FMRP were highly successful in FMR1 knockout mouse models.

This Special Issue will comprise reviews and original research articles focused on the recent advances in genetics/genomics testing; the contribution of mosaicism and epigenetic processes; and the clinical description, co-morbidities, biomarkers, and natural history of FXS. Current and future directions with a focus on improved screening, diagnosis, and treatment will be addressed in this issue.

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Message from the Editor-in-Chief

Genes are central to our understanding of biology, and modern advances such as genomics and genome editing have maintained genetics as a vibrant, diverse and fastmoving field. There is a need for good quality, open access journals in this area, and the *Genes* team aims to provide expert manuscript handling, serious peer review, and rapid publication across the whole discipline of genetics. Starting in 2010, the journal is now well established and recognised.

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