



## Diagnosis of Neurogenetic Disorders: Contribution of Next Generation Sequencing and Deep Phenotyping

Guest Editor:

**Dr. Alisdair McNeill**

Sheffield Institute for  
Translational Neuroscience,  
Department of Neuroscience,  
University of Sheffield, 385a  
Glossop Road, Sheffield, S10  
2HQ, UK

Deadline for manuscript  
submissions:

**closed (1 February 2019)**

### Message from the Guest Editor

The contribution of genomic variants to the aetiopathogenesis of both paediatric and adult neurological disease is increasingly recognised. The use of next generation sequencing has led to the discovery of novel neurodevelopmental disorders, and provided insight into the aetiopathogenesis of common adult neurological diseases. Correctly classifying the pathogenicity of genomic variants from amongst the large number of variants identified by next generation sequencing is recognised as perhaps the major challenge facing the field. Deep phenotyping techniques can aid variant interpretation by correctly classifying individuals as affected or unaffected for segregation studies. The lack of information on the clinical phenotype of novel genetic subtypes of neurological disease creates limitations for Genetic Counselling. Both deep phenotyping and qualitative studies can capture the clinical and patient's perspective on a disease and provide valuable information.





an Open Access Journal by MDPI

## Editor-in-Chief

### Prof. Dr. Stephen D. Meriney

Department of Neuroscience,  
University of Pittsburgh,  
Pittsburgh, PA 15260, USA

## Message from the Editor-in-Chief

You are invited to contribute a research article or a comprehensive review for consideration and publication in *Brain Sciences* (ISSN 2076-3425). *Brain Sciences* is an open access, peer-reviewed scientific journal that publishes original articles, critical reviews, research notes, and short communications on neuroscience. The scientific community and the general public can access the content free of charge as soon as it is published.

## Author Benefits

**Open Access:** free for readers, with article processing charges (APC) paid by authors or their institutions.

**High Visibility:** indexed within Scopus, SCIE (Web of Science), PubMed, PMC, Embase, PSYINDEX, CAPlus / SciFinder, and other databases.

**Rapid Publication:** manuscripts are peer-reviewed and a first decision is provided to authors approximately 12.9 days after submission; acceptance to publication is undertaken in 2.5 days (median values for papers published in this journal in the first half of 2024).

## Contact Us

---

Brain Sciences Editorial Office  
MDPI, Grosspeteranlage 5  
4052 Basel, Switzerland

Tel: +41 61 683 77 34  
[www.mdpi.com](http://www.mdpi.com)

[mdpi.com/journal/brainsci](https://mdpi.com/journal/brainsci)  
[brainsci@mdpi.com](mailto:brainsci@mdpi.com)  
[X@BrainSci\\_MDPI](https://twitter.com/BrainSci_MDPI)