

Editorial

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# Translational aspects in neurodevelopmental disorders

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The past decade has impressively increased the diagnostic possibilities and outcomes for neurodevelopmental disorders (NDDs)<sup>[1-5]</sup>. Therefore, the scope of this Special Issue “Translational aspects in neurodevelopmental disorders” is broadened from gene and syndrome discovery to the clinical and biological understanding of defects underlying the NDDs. As over 50% of underlying genetic defects can now be identified in individuals with congenital neurodevelopmental problems, this provides challenges and opportunities for novel trajectories in clinical, molecular, and (fundamental) neurobiology fields.

Excellent studies that cover this broad scope have been peer reviewed and selected for publication in this Special Issue. Such studies are highly relevant to move this field forward from disorder causation to the understanding and intervention strategies on various aspects. The manuscript “Intellectual disability, the long way from genes to biological mechanisms”<sup>[6]</sup> describes several aspects in this broad trajectory. More classical genotype-phenotype studies are shown in two examples: “Spectrum of MECP2 mutations in Indian females with Rett Syndrome - a large cohort study” and “Assigning single clinical features to their disease-locus in large deletions: the example of chromosome 1q23-25 deletion syndrome”<sup>[7,8]</sup>.

In addition to genomic technologies, important contributions to brain functioning can be provided by imaging modalities. The development and application of technologies that can explore important aspects of the anatomy, physiology, or biochemistry of the central nervous system are given and their properties, as well as pros and cons, are discussed in the context of several inborn errors of metabolism<sup>[9]</sup>.



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Tools that further define clinical and molecular phenotypes and may assist in molecular confirmation are shown by the use of facial recognition in “DeepGestalt analysis of the *SETD5*-associated intellectual disability syndrome”<sup>[10]</sup> and by the application of DNA episigns in the study on several molecular variants causing Kleefstra syndrome in “*EHMT1* pathogenic variants and 9q34.3 microdeletions share altered DNA methylation patterns in patients with Kleefstra syndrome”<sup>[11]</sup>.

The involvement of molecular and biological pathways involved in NDDs are reflected nicely by two different reviews: “Role of transfer RNA modification and aminoacylation in the etiology of congenital intellectual disability” and “Chromodomain helicase DNA-binding proteins and neurodevelopmental disorders”<sup>[12,13]</sup>. The NDDs field increasingly benefits from the use of cellular and animal models, as reflected by the studies on “Human induced pluripotent cells in personalized treatment of monogenic epilepsies”<sup>[14]</sup> and “Animal models of autism: a perspective from autophagy mechanisms”<sup>[15]</sup>. Finally, the manuscript “The influence of CYP enzymes and ABCB1 on treatment outcomes in schizophrenia: association of CYP1A2 activity with adverse effects” highlights the importance of pharmacogenomics on treatment of developmental or psychiatric conditions<sup>[16]</sup>.

Altogether, the studies presented in this special issue highlight the fascinating and rapidly moving field on understanding and treating Mendelian NDDs. Unique strategies are developed for optimal personalized medicine targeted to this vulnerable population.

## DECLARATIONS

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The author contributed solely to the article.

### Availability of data and materials

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### Conflicts of interest

The author declared that there are no conflicts of interest.

### Ethical approval and consent to participate

Not applicable.

### Consent for publication

Not applicable.

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