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**Investigating Genes, Environments, and their Interactions in the Service
of Informing Individualized Diagnosis and Treatment in Developmental
Disabilities**

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Investigating Genes, Environments, and their Interactions in the Service of Informing Individualized Diagnosis and Treatment in Developmental Disabilities

Developmental outcomes are determined by the interactions between myriad factors operating at multiple different levels (Bronfenbrenner, 1992; Sameroff, 2009). Broadly speaking, these factors may be related to the genetic background of the individual (genotype) as well as from the external world (environment) with which the individual interacts. During development, the interaction between genotype and environment (Genotype x Environment or G×E) is paramount to the ontogeny of the specific phenotype, which is the set of observable characteristics of the individual. Indeed, one way to conceptualize the diverse array of resulting phenotypes is as the result of different genotypes responding to environmental variation in different ways.

Although there is a long tradition of studies on GxE in psychiatry (for example on schizophrenia and depression), researchers working within field of developmental disabilities have been slower to adopt this methodology. Thus, the field of developmental psychopathology has not yet reached the maturity of being able to use science to inform individualized diagnosis and treatment. And yet, though conclusive indicators of causal GxE patterns are largely lacking in the field, the results obtained so far in this discipline highlight the need to adopt a multilevel approach to understanding the development of psychopathology, one that combines genetic, environmental, epigenetic, neural, and hormonal perspectives, in the service of better understanding the mechanisms governing the ontogeny and outcome of different developmental disabilities. Understanding GxE patterns that confer

risk for developmental disabilities might enable practitioners to design customized treatment options for specific subgroups of patients.

For this special issue, we have collected empirical studies as well as review and perspective papers that focus on gene and environmental factors, as well as their interplay, in understanding risk for developmental disabilities (i.e., any of the chronic developmental conditions described in the DSM-5).

The current special issue begins with a conceptual analysis of the meaning and interpretation of Genotype $\times$ Environment interactions (Moore, 2018) and the specific roles of specific genes and environments as risk factors for the development of specific conditions such as ADHD (Azaredo et al. 2018), Autism Spectrum Disorders (Doi et al., 2018), or anxiety disorders (Boparai et al., 2018). Then, we collected articles that focus on the effect of environment in the forms of early caregiving experiences (Cataldo et al., 2018; Partington et al. 2018), or early maltreatment (Maglione et al. 2018; Torrisi et al. 2018) and their effects on child development. Braam and colleagues (2018) assess how other environmental factors, specifically maternal melatonin levels (Braam et al. 2018) and gut microbiota (Zhou et al. 2018), modulate the risk for autism spectrum disorder risk in children. In an attempt to present a well-rounded view of Gene $\times$ Environment interactions, we also have selected articles focusing on the impacts of the individual on the environments in which they are embedded. One paper (Zhang et al., 2018) focuses on how children with Rett Syndrome respond to their names, offering the suggestion that this may be an early biomarker of the disorder. This special issue contains a prominent focus on early linguistic environment, with four articles exploring different aspects of linguistic experience and its

interaction with genetic profiles, ranging from the study of language in Rett Disorder (Pokorny et al., 2018), language development (Onnis et al., 2018), dyslexia (Mascheretti, Andreola, Scaini, & Sulpizio, 2018), and dyadic matching of language styles (Boporai et al., 2018). The special issue concludes with an article in which Zhou, Mc Adam and Donnelly (2018) introduce the concept of endophenotypes presenting a link between Anorexia and ASD.

In assembling this special issue, we hope we have called attention to an area of research in which promising discoveries are on the horizon, as well as that we have stimulated interest in the investigation of Gene x Environment interactions in diagnosing as well as in predicting outcomes for children with developmental disabilities.

References

- Azeredo, A., Moreira, D., & Barbosa, F. (2018). ADHD, CD, and ODD: Systematic review of genetic and environmental risk factors. *Research in Developmental Disabilities*.
<https://doi.org/10.1016/j.ridd.2017.12.010>
- Bronfenbrenner, U. (1992). Ecological systems theory. In R. Vasta (Ed.), *Six theories of child development: Revised formulations and current issues* (pp. 187-249). London, England: Jessica Kingsley Publishers.
- Boparai, S., Borelli, J. L., Partington, L., Smiley, P., Jarvik, E., Rasmussen, H. F., ... & Nurmi, E. L. (2018). Interaction between the Opioid Receptor OPRM1 Gene and Mother-Child Language Style Matching Prospectively Predicts Children's Separation Anxiety Disorder Symptoms. *Research in Developmental Disabilities*.
<https://doi.org/10.1016/j.ridd.2018.03.002>
- Braam, W., Ehrhart, F., Maas, A.P.H.M., Smits, M.G., Curfs, L. (2018). Low maternal melatonin level increases autism spectrum disorder risk in children. *Research in Developmental Disabilities*. <https://doi.org/10.1016/j.ridd.2018.02.017>
- Cataldo, I., Azhari, A., Lepri, B., & Esposito, G. (2017). Oxytocin receptors (OXTR) and early parental care: An interaction that modulates psychiatric disorders. *Research in Developmental Disabilities*. <https://doi.org/10.1016/j.ridd.2017.10.007>
- Doi, H., Fujisawa, T. X., Iwanaga, R., Matsuzaki, J., Kawasaki, C., Tochigi, M., ... & Shinohara, K. (2018). Association between single nucleotide polymorphisms in estrogen receptor 1/2 genes and symptomatic severity of autism spectrum disorder. *Research in Developmental Disabilities*. <https://doi.org/10.1016/j.ridd.2018.02.014>
- Mascheretti, S., Andreola, C., Scaini, S., & Sulpizio, S. (2018). Beyond genes: A systematic review of environmental risk factors in specific reading disorder. *Research in Developmental Disabilities*. <https://doi.org/10.1016/j.ridd.2018.03.005>
- Moore, D. S. (2018). Genex Environment interaction: What exactly are we talking about?. *Research in Developmental Disabilities*. <https://doi.org/10.1016/j.ridd.2018.04.012>
- Onnis, L., Truzzi, A., Ma, X. (2018). Language development and disorders: Possible genes and environment interactions. *Research in Developmental Disabilities*.
<https://doi.org/10.1016/j.ridd.2018.06.015>

- Partington, L. C., Borelli, J. L., Smiley, P., Jarvik, E., Rasmussen, H. F., Seaman, L. C., & Nurmi, E. L. (2018). Parental overcontrol x OPRM1 genotype interaction predicts school-aged children's sympathetic nervous system activation in response to performance challenge. *Research in Developmental Disabilities*.
<https://doi.org/10.1016/j.ridd.2018.04.011>
- Pokorny, F. B., Bartl-Pokorny, K. D., Einspieler, C., Zhang, D., Vollmann, R., Bölte, S., et al. (2018). Typical vs. atypical: Combining auditory Gestalt perception and acoustic analysis of early vocalisations in Rett syndrome. *Research in Developmental Disabilities*. <https://doi.org/10.1016/j.ridd.2018.02.019>
- Sameroff, A. (2009). The transactional model. In A. Sameroff (Ed.), *The transactional model of development: How children and contexts shape each other* (pp. 3-21). Washington, DC, US: American Psychological Association.
<http://dx.doi.org/10.1037/11877-001>
- Torrìsi, R., Arnautovic, E., Perizzolo, V. P., Vital, M., Manini, A., Suardi, F., et al. (2018). Developmental delay in communication among toddlers and its relationship to caregiving behavior among violence-exposed, posttraumatically stressed mothers. *Research in Developmental Disabilities*. <https://doi.org/10.1016/j.ridd.2018.04.008>
- Zhang, D., Roche, L., Bartl-Pokorny, K. D., Kriebler, M., McLay, L., Bölte, S., ... & Marschik, P. B. (2018). Response to name and its value for the early detection of developmental disorders: Insights from autism spectrum disorder, Rett syndrome, and fragile X syndrome. A perspectives paper. *Research in Developmental Disabilities*.
<https://doi.org/10.1016/j.ridd.2018.04.004>
- Zhou, Z. C., McAdam, D. B., & Donnelly, D. R. (2017). Endophenotypes: A conceptual link between anorexia nervosa and autism spectrum disorder. *Research in Developmental Disabilities*. <https://doi.org/10.1016/j.ridd.2017.11.008>
- Zhou, J., He, F., Yang, F., Yang, Z., Xie, Y., Zhou, S., ... & Zhou, W. (2017). Increased stool immunoglobulin A level in children with autism spectrum disorders. *Research in Developmental Disabilities*. <https://doi.org/10.1016/j.ridd.2017.10.009>