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Disorders of Transcriptional Regulation

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Deadline for manuscript submissions:

closed (30 April 2020)

Message from the Guest Editors

DTR include a growing list of monogenic disorders that overlap phenotypically and mechanistically due to direct and indirect effects of mutant alleles on gene transcription and regulation. Examples include Kabuki syndrome, CHARGE syndrome in addition to many others. These disorders share many unmet needs, including a significant disease burden with frequent multiorgan involvement, limited knowledge on disease progression and genotypephenotype correlation, and lack of biomarkers and therapies. The broad use of next-generation sequencing has not only resulted in an increased diagnostic rate but also the identification of novel disease genes that affect transcriptional regulation. Recent breakthroughs have led to a deeper understanding of disease-related mechanisms that may result in the identification of therapeutic targets to reverse aspects of the clinical phenotype. We extend an invitation for original research and review articles that focus on clinical, molecular genetic or therapeutic aspects involving any of the DTRs.













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Message from the Editor-in-Chief

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