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Editorial: Translational Science of Rare Diseases

After seven years since this journal first initiated its publication, it is appropriate to review what has occurred since that time and outline our strategies going forward. This is occasioned by the change in executive leadership. *Translational Science of Rare Diseases* (TSRD) was initiated to fill a void in scientific literature related to the translational approaches to rare diseases. Bridging the interface between basic and clinical research has become increasingly important as the development of new therapies for clinical disease is critical to the advancement of care for medical problems, most especially for the rare diseases that have lacked specific treatments.

TSRD has succeeded in its early development in providing a voice for disorders which affect a relatively small number of individuals, in the US less than one in 200,000 individuals or in the EU less than one in 2,000 individuals. Still, these efforts have lacked sufficient currency to elevate the journal to a proper recognition in relevant databases such as *PubMed*, *Scopus*, and *Web of Science*. It is our intention over the next few years to elevate this position by increasing our publication record and expanding our readership. We continue to feature independent, original research articles as well as in depth review of specific rare diseases and therapeutics, and opinion pieces from regulators, patient advocates, industry and academic researchers focused on rare diseases.

This posture is in line with recent changes in the administrative leadership of TSRD with the addition of Wilbert van der Sluijs as managing publisher and Marleon Dias Jones in journal promotion. With their assistance we have updated our operational and journal guidelines in keeping with current requirements and accepted practices.

As these are now advancing, we would invite you to view the contents of the current issue. These include what is involved in becoming a research participant to help all navigate what may be unfamiliar territory. We, also, present new information on ciliopathy gene variants in infants in relation to congenital heart disease and heterotaxy.

Finally, the current issue concludes with a comparison of research efforts in four neurodevelopmental disorders: Rett syndrome, CDKL5 Deficiency Disorder, FOXG1 disorders, and MECP2 Duplication Disorder. These disorders are described individually and then collectively based on efforts at biomarker development.

We believe you will find something of interest in this issue of TSRD and enjoy the advances we are making to elevate the journal to the next level.

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