Editor's note:
"Rare Diseases Column" is chaired by Dr. Zhanhe Wu from The Children's Hospital at Westmead, Australia, featuring articles related to rare diseases mostly genetic based, presented in early life disease, with chronic phase but frequently progressive, disabling and life threatening diseases. Article types of original articles, review articles, case reports, perspectives, etc. are welcomed to be submitted to the column.

Rare Diseases Column (Editorial)

How to make the timely diagnosis to stabilize disease progression and prevent the organ damage from Fabry disease?

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It is estimated that there are more than 8,000 different types of rare diseases affecting fewer than 1 in 2000 people although the exact number remains to be calculated. Research into rare diseases in the understanding of rare diseases mechanisms, leading to potential new treatments has been interested strongly to improved targeted and personalised approaches to healthcare.

In the column of rare disease of this issue, we are very honoured to invite an expert in genetic metabolic disease, Dr Carolyn Ellaway from the Western Sydney Genetics Programs, Children's Hospital at Westmead, Australia to contribute a special article titled "Paediatric Fabry disease".

As Dr Ellaway summarised that Fabry disease is a rare, progressive X-linked inborn error of the glycosphingolipid metabolic pathway. Mutations of the *GLA* gene result in deficiency of the lysosomal enzyme, α-galactosidase A with accumulation of glycosphingolipids, particularly globotriaosylceramide (GL3) in the vascular endothelium of various tissues.

In this article, Dr Ellaway has systemically described the pathophysiology, epidemiology, clinical manifestations, diagnosis, importance of early diagnosis, management and treatment of the Fabry disease. If you are interested in on how the timely diagnosis can be made, please read Dr Ellaway's clear, comprehensive and advanced article "Paediatric Fabry disease".

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Footnote

Conflicts of Interest: The author has no conflicts of interest to declare.

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