EDITORIAL

Next-generation sequencing applied in pediatric diseases

Next-generation sequencing (NGS) has been widely conducted in clinical testing for human diseases, and a series of bioinformatics pipelines have been developed to analyze the resulting large data. However, most testing or research focused on adult diseases, while a few cared about pediatric diseases. Considering the different etiology and development of pediatric diseases, compared to the adult diseases, it is necessary to revaluate or modify the standard bioinformatics pipelines for NGS data treatment in clinical testing for pediatric diseases.

Jin and her colleagues performed an excellent job that not only summarized the common pipelines for NGS data, but also provided new suggestions for bioinformatical methods applied in clinical testing for pediatric disease, including rare genetic diseases, rare sporadic diseases and familial pediatric tumors.[1] For rare genetic diseases, family-based NGS analysis combining with clinical information exhibits particular importance, due to the atypical symptoms. To identify novel pathogenic mutation resulting in the pediatric rare sporadic diseases, at least two probands should be recruited. As to familial pediatric tumors, NGS for germline DNA samples from family members should be recommended to identify new pathogenic variants, using flexible genetic models. All the evidences mentioned above showed that the bioinformatical pipelines applied in clinical testing for pediatric diseases are variable, and might be modified based on the etiology and symptom of the diseases.

Considering known gene panels is insufficient to detect pathogenic genes or genetic variants for pediatric diseases, seeking new biomarkers using NGS in clinical testing becomes an urgent project in pediatric medicine. On our wish list, the children might benefit from NGS testing in diagnosis as soon as possible, however the methodology and genetic models for data analysis have not been standardized till now, and the available methods still need to be validated in more patients.

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CONFLICT OF INTEREST

I declare that they I no competing interests.

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