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Closing the diganostic gap in rare genetic conditions

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Abstract

The diagnosis of rare genetic diseases has seen a leap in recent years thanks to technological advances in next generation sequencing. Government and private initiatives facilitate the adoption of such technologies both in clinical and research settings, which has advanced our understanding of rare genetic conditions. However, the diagnostic yield (30-60%) reflects the need for further approcahes to address this diagnostic gap. Lack of diagnosis is problematic at all levels and is mainly translated into missed opprtunities of coordinated care and potential therapies. Therore, despite the promissing achievements in both diagnosis methodologies and novel therapies, many patients remain undiagnosed. To address this unmet clinical need, we put together a framework that can easily be adopted in clinical and research seetings to narrow the diagnosic gap. This diagnotic gap workflow is a multidisciplinary approach based on data sharing, data mining, functional work and and up to date biobase. Here we explain the diagnostic gap workflow, and give an example of how it has led to improved diagnostic rate in a number of critically ill infants who remained without a diagnosis despite having had whole genome sequencing. We propose this workflow can seemlessly be implemented without the need for sophsticated infrastructure.

Biography

Lamia Mestek Boukhibar a BscHons degree in molecular genetics from King's College London (2007) and completed her DPhil in genetics at Oxford University in 2010. She has held several postdoctoral positions at University College London and Imperial College where she worked on several basic research projects addressing the genetics of healthy ageing, and the genetics of biological robustness. Lamia then moved from basic to translational research in 2016 when she joined the Great Ormond Street Institute of Child Health and Great Ormond Street Hospital. Lamia put her experience in genetics and genomics to address unmet clinical needs where she led the reserach side of developing a robust workflow to offer rapid whole genomne sequening for critically ill infants with a suspected rare genetic condition. She has now focused her interest in addressing yet another unmet clinical need which is the diagnostic odysssey. She has implemneted simple yet effecttive workflow to close the diagnostic gap. She published in several peer reviewed journals and was invited to several national and international conferences and seminars.

Publications

1. Bi-allelic Variants in TKFC Encoding Triokinase/FMN Cyclase Are Associated with Cataracts and Multisystem Disease, January 2020The American Journal of Human Genetics 106(2)
2. Sotos Syndrome Presenting with Neonatal Hyperinsulinaemic Hypoglycaemia, Extensive Thrombosis, and Multisystem Involvement, March 2019Hormon Research in Pediatrics 92(1):1-7, DOI:10.1159/000496545
3. Making a rapid genetic diagnosis in a neonate with anuric renal failure, October 2018

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