NGS in diagnostics - where things can go wrong

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August 2, 2021

Abstract

Massive parallel sequencing technology has become the predominant technique for genetic diagnostics and research. Many genetic laboratories have wrestled with the challenges of setting up genetic testing workflows based on a completely new technology. The learning curve we went through as a laboratory was accompanied by growing pains while we gained new knowledge and expertise. Here we discuss some important mistakes that have been made in our laboratory through ten years of clinical exome sequencing but that have given us important new insights on how to adapt our working methods. By providing these examples and the lessons that we learned from them, we hope that other laboratories do not need to make the same mistakes.

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