QUALITY ASSURANCE OF HIGH THROUGHPUT SEQUENCING IN THE DIAGNOSTIC LABORATORY

Gary Van Domselaar, Ph.D.

Chief Bioinformatics, National Microbiology Laboratory,
Public Health Agency of Canada, Canadian Science Centre for Human and Animal Health,
Winnipeg, Canada R3E 3R2
Chef Bioinformatique, Laboratoire National de Microbiologie, Agence de la Santé Publique du Canada, Centre scientifique Canadien de la Santé Humaine et Animale, Winnipeg, Canada R3E 3R2
gary.vandomselaar@phac-aspc.gc.ca
Telephone | Téléphone 204-784-5994/ Facsimile | Télécopieur 204-784-7546

Modern sequencing technologies and associated bioinformatics analysis pipelines have revolutionised the biological research landscape. However, translating these technologies out of the research laboratory and into routine use for diagnostic applications has been slow. This is due, in part, to the disruptive nature of the technology requiring the rethinking and development of novel analysis methodologies including notably, big data analytics. Quality control and quality assurance of these vast, imperfect data sets has also been slow as the communities of expertise qualified to define the best practices and standards for the application of high throughput sequencing for many diagnostic applications are only now being assembled. In this talk I will discuss these challenges and efforts to address them from the perspective of a Canadian Federal bioinformatics scientist, with a focus on quality assurance and quality control for whole genome sequence-based disease surveillance, microbial typing, and microbial forensics.