Extended Abstract
Volume 4:1, 2020

Research and Reports in Medical Sciences

Open Access

Molecular pathology 2020: Challenges of Molecular Pathology

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Abstract

Keywords:
overdiagnosis; overtreatment; CBBLE (case-based-based blended learning ecosystem);case studies; precision medicine; omics driven; low resource setting; high resource setting

Introduction:
The revolutionary advances made in molecular biology are occurring in such a rate that it is drastically changing the methods of diagnosis of disease. However practicing as a pathologist in Kathmandu University Hospital (KUH), we have huge limitations.
Precision medicine aims to focus on meeting patient requirements accurately,optimizing patient outcomes, and reducing under-/overdiagnosis and therapy. We aim to offera fresh perspective on accuracy driven "age-old precision medicine" and illustrate how newercase-based blended learning ecosystems (CBBLE) can strengthen the bridge between age-old precisionapproaches with modern technology and omics-driven approaches. Methodology: We present aseries of cases and examine the role of precision medicine within a "case-based blended learningecosystem" (CBBLE) as a practicable tool to reduce overdiagnosis and overtreatment. We illustratedthe workflow of our CBBLE through case-based narratives from global students of CBBLE in highand low resource settings as is reflected in global health. Results: Four micro-narratives based oncollective past experiences were generated to explain concepts of age-old patient-centered scientificaccuracy and precision and four macro-narratives were collected from individual learners in ourCBBLE. Insights gathered from a critical appraisal and thematic analysis of the narratives werediscussed. Discussion and conclusion: Case-based narratives from the individual learners in ourCBBLE amply illustrate their journeys beginning with "age-old precision thinking" in low-resourcesettings and progressing to "omics-driven" high-resource precision medicine setups to demonstratehow the approaches, used judiciously, might reduce the current pandemic of over-/underdiagnosisand over-/undertreatment.

The term "Precision Medicine" was first coined by Clayton Christensen in his book the"Innovator's Prescription", published in 2009 [1]. According to the early definition given by theInstitute of Precision Medicine, "Precision medicine is targeted, individualized care that is tailored toeach patient based on his or her specific genetic profile and medical history" [2].While the above definitions allow us to assume that precision medicine is focused on meetingpatients' requirements accurately, we need to review the scientific nature of accuracy, precision andtheir relationship with each other to put things in perspective. This is essential toward optimizingpatient requirements and outcomes, minimizing damage to the healthcare ecosystem by reducingunder-overdiagnosis and therapy. To quote from Thomas (2014), "The healthcare 'system' is nowbetter understood as an 'ecosystem' of interconnected stakeholders, each one charged with a missionto improve the quality of care while lowering its cost. To ensure patient safety and quality care whilerealizing savings, these stakeholders are building new relationships—often outside the four walls ofthe hospital" [3].
We illustrate the above concepts with micro case studies below:"An elderly patient from a country endemic with tuberculosis presented with a chronic cough andweight loss. A lung pathology was detected on imaging that was not amenable to further biopsyefforts as a result of unavailable resources. He was started on empirical treatment for tuberculosisafter sending a sputum for acid-fast bacillus (AFB) and culture."In tuberculosis endemic countries, physicians often treat empirically for tuberculosis in suspiciouslung pathologies, although lung malignancy is a close differential in such situations. In the abovepatient's context, physicians were being obviously imprecise in starting treatment for tuberculosisempirically even when the tuberculosis bacilli was undetectable. This is an acceptable standardpractice with established protocols for treating sputum-negative tuberculosis utilized globally by manycountries that are endemic for tuberculosi

"The idea of sharing and learning around patients has been alive since the beginning of medicine,when physicians would present their cases to a large audience to primarily learn from the inputs ofother physicians." With the invention of the printing press, instead of restricting themselves to verbalface-to-face case presentations, many physicians published their cases in journals and slowly the medicalfraternity started naming those published diseases after their first authors. "In this way, case reportingbecame a gainful activity not only in terms of scientific advancement towards patient benefits, but alsoas an important instrument of physician fame." We have utilized this case reporting model to help ourpatients and to train our medical students about disease and patient experience
Healthcare 2018,6, 78 5 of 17By reporting cases, this model allows more engagement both from patients and medical studentsto reach a precise and accurate diagnosis, and also helps as an educational tool.

Objectives:
To study the molecular tests performed in KUH, the challenges faced and to explore the additional possibilities in the field of Molecular pathology.
Materials and methods: The cases and problems faced in Molecular tests by the doctors, and technicians are discussed, analysed and reported.
Results: The molecular tests currently being performed in KUH are PCR and Immunohistochemistry (IHC). PCR test functioning since 1 year is performed especially for HPV detection and for diagnosing Tuberculosis. Total PCR cases sent are average 10 per day. Among 580 cancer case in a year, 34 needed IHC for conformation. Only 4 are performed IHC.

Discussion:
Use of Molecular pathology tests is at its infancy in KUH. IHC is very difficult to continue because of the cost and hence the scarcity of the cases. The antibody once opened has very short life that aids in increasing the cost. So the patients who can afford are sent abroad. However Kathmandu University have nine affiliated colleges. If we can have standard Molecular diagnosis lab then we can collect the cases from almost all of them which can reduce the cost.
Conclusion: There's frequent diagnosis of cancer cases in KUH and in affiliated colleges, KUH can be made the hub for Molecular diagnosis in Nepal.

This work is partly presenting at 2nd International Conference on Molecular Pathology and Genomics on December 09-10, 2020