DNA sequencing

DNA sequencing is the process of determining the nucleic acid sequence – the order of nucleotides in DNA. It includes any method or technology that is used to determine the order of the four bases: adenine, guanine, cytosine, and thymine.

Genomics is a discipline in genetics that applies recombinant DNA, DNA sequencing methods, and bioinformatics to sequence, assemble, and analyze the function and structure of genomes.

MI GPSai, a Genomic Prevalence Score, uses DNA sequencing and whole transcriptome sequencing data coupled with machine learning to aid in the diagnosis of cancer. The algorithm trained on genomic data from 34,352 cases and genomic and transcriptomic data from 23,137 cases and was validated on 19,555 cases. MI GPSai predicted the tumor type in the labeled data set with an accuracy of over 94% on 93% of cases while deliberating amongst 21 possible categories of cancer. When also considering the second highest prediction, the accuracy increases to 97%. Additionally, MI GPSai rendered a prediction for 71.7% of CUP cases. Pathologist evaluation of discrepancies between submitted diagnosis and MI GPSai predictions resulted in change of diagnosis in 41.3% of the time. MI GPSai provides clinically meaningful information in a large proportion of CUP cases and inclusion of MI GPSai in clinical routine could improve diagnostic fidelity. Moreover, all genomic markers essential for therapy selection are assessed in this assay, maximizing the clinical utility for patients within a single test [1)].

Bächli et al., report a single-institutional collection of pediatric brain tumor cases that underwent a refinement or a change of diagnosis after completion of molecular diagnostics that affected clinical decision-making including the application of molecularly informed targeted therapies. 13 pediatric central nervous system tumors were analyzed by conventional histology, immunohistochemistry, and molecular diagnostics including DNA methylation profiling in 12 cases, DNA sequencing in 8 cases and RNA sequencing in 3 cases. 3 tumors had a refinement of diagnosis upon molecular testing, and 6 tumors underwent a change of diagnosis. Targeted therapy was initiated in 5 cases. An underlying cancer predisposition syndrome was detected in 5 cases. Although this case series, retrospective and not population based, has its limitations, insight can be gained regarding precision of diagnosis and clinical management of the patients in selected cases. Accuracy of diagnosis was improved in the cases presented here by the addition of molecular diagnostics, impacting clinical management of affected patients, both in the first-line as well as in the follow-up setting. This additional information may support the clinical decision making in the treatment of challenging pediatric CNS tumors. Prospective testing of the clinical value of molecular diagnostics is currently underway [2)].

1)
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2)
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