Clinical applications of DNA sequencing

Reports of genomic sequencing being applied to the medical care of individual patients have begun to appear in the scientific literature, but much needs to be done before the use of sequence data will be routine. It is unclear what diseases or individual susceptibilities will be usefully addressed by a genomic sequencing approach, or the potential range of clinical applications. Furthermore, incorporation of comprehensive genomic sequence data into clinical care will require changes to institutional policies, standard procedures (including simplified analysis and interpretive tools), and the ability to integrate sequence information into the clinical workflow. The ethical and psychosocial implications of returning genomic variation data, with all of the caveats regarding statistical uncertainties and incomplete knowledge, remain poorly understood.

NYUMC has a Genome Technology Center with the latest DNA sequencing technology (currently the Illumina Hiseq 2000) and a bioinformatics group dedicated to analyzing this data. All of our current projects are research oriented, but some are also explorations of clinical translational applications. We are particularly interested in developing diagnostic sequencing for cancer patients using the Exome sequencing approach.

NYUMC is developing a program to bring together genomics specialists, clinicians, and bioinformaticians to study the challenges of utilizing genomic sequence data in the clinic in the routine practice of medicine. The challenges are many and not disease-specific. Possible areas for medical student research include: development of technical specifications and standards for sequencing in a clinical setting, creating standards of clinical significance for observed sequence variants, and investigation of methods to transmit genome-scale data to physicians in a fashion and timescale that fits the normal clinical workflow.

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