Genotyping and Anatomical Abnormalities

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The advances in human genetics during the past three decades have resulted from a series of technological and organizational breakthroughs. Among these is the development of accurate sequencing technologies that allowed the determination of the human genome sequence. The multinational consortium that produced that sequence then went on to determine variation among people of different ethnicities. In turn, the database of variation from sequencing was coupled with new technologies for assaying variation in tens of thousands of individuals and at increasingly greater densities. These genotyping arrays are now being used to identify DNA variation associated with risk for disease and to discover new genes that may represent targets for pharmaceutical interventions. As the limits of genotyping are realized a renewed emphasis on cheaper and faster sequencing approaches has emerged and various new and exciting approaches to whole genome sequencing are fast appearing. This talk will outline some of the technologies that have been and are being developed to increase the speed and accuracy of genetic data and how such information will revolutionize the way medicine is practiced for the rest of the century.