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In 2004, the Chromosome 18 Registry & Research Society, along with several other advocacy organizations, convened the first ever World Congress on Chromosome Abnormalities. Two groups of people participated in this meeting: people with an interest in different chromosome abnormalities and experts from various medical fields. During this meeting, discussions focused on the effects of changes in gene copy number as well as the importance of completely characterizing those medical and developmental effects.
Since this meeting, technological developments have allowed scientists to see that there is much more variation in the human genome than originally thought. The challenge now is to understand which of those changes lead to medical and developmental problems. This manuscript proposed some ways to classify and determine the effects of a deletion or duplication of a specific gene. The ultimate goal of these efforts will be to develop a genome-wide gene dosage map. This map will make it possible for physicians to look at a person‟s genetic test result, determine which genes will result in medical or developmental issues, and treat accordingly.