



**FIGURE 13.32.** There is extensive variation in gene content within the human population. DNA from 20 individuals revealed genome-wide variation in copy number at 76 loci, which reflects the presence of deletions and duplications. These polymorphisms each average 465 kb in length and contain 70 known genes in total. Some polymorphisms coincide with loci that have a high rate of chromosomal rearrangement, causing inherited diseases (A, Prader–Willi and Angelman syndromes; B, cat eye syndrome; C, DiGeorge/velocardiofacial syndrome; D, spinal muscular atrophy). This survey should detect most large-scale deletions and insertions but will miss smaller rearrangements.

13.32, modified from Sebat J. et al., *Science* **305**: 525–528, © 2004 American Association for the Advancement of Science