

Chromosome Analysis: Banding Patterns and Structural Aberrations

David F. Holland, M.S., *The University of Texas Southwestern Medical Center at Dallas*

Purpose

Students will use ideograms of chromosomes from four primates (human, chimpanzee, gorilla, and orangutan) to investigate differences in banding patterns between the species. From this information they will determine which types of chromosomal aberrations (deletions, translocations, etc.) may have taken place to give rise to these differences.

Materials

Colored Pencils or Markers

Background

To prepare chromosomes for analysis, a culture of rapidly dividing cells must be obtained. This is most usually achieved by drawing a blood sample, which is centrifuged to separate the white and red cells. The white cells are then cultured in a medium containing a substance that stimulates mitosis. After 72 hours of incubation, colchicine is added to “freeze” the cells in metaphase of mitosis. A hypotonic solution is then added to swell the cells and separate the chromatids. The cells are then fixed, spread on slides and stained. The Giemsa stain produces light and dark bands on the chromosomes, which are used in describing the structure of chromosomes. The chromosomes are photographed, cut from the photomicrograph and arranged in pairs. This process is called karyotyping. A typical chromosome spread and karyotype are shown in Figure 1. By examining chromosomes in this way, many abnormalities may be identified.

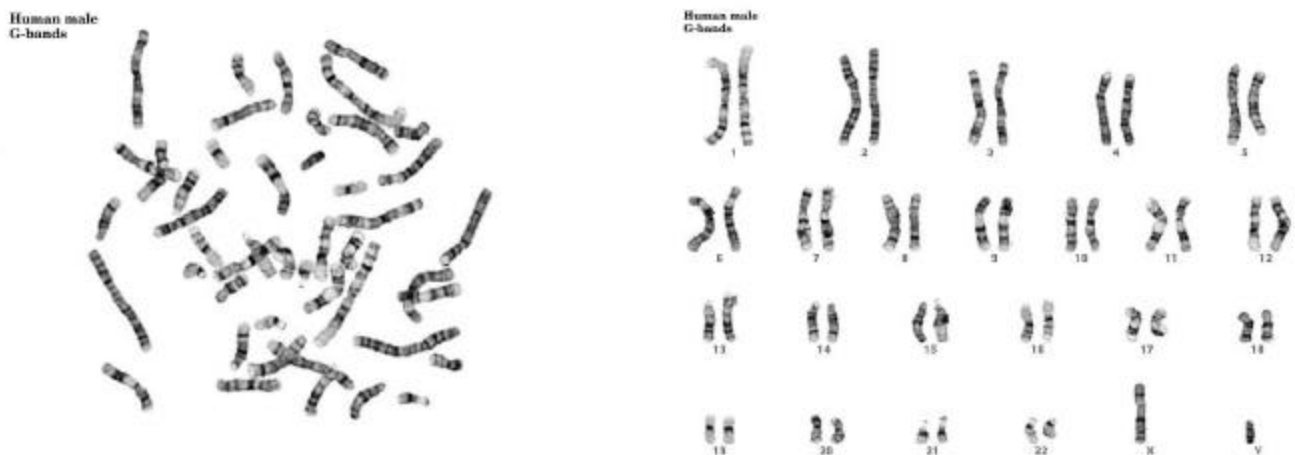
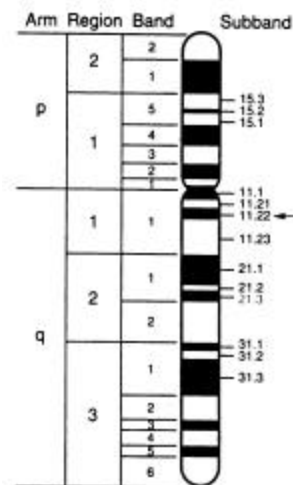


Figure 1 (left) A chromosome spread from a lymphocyte culture using Giemsa staining. (right) Normal male karyotype.

Chromosomes are composed of supercoiled DNA and its associated proteins. This material is called chromatin. Chromosomes consist of two sister chromatids connected at a constricted region called the centromere. The centromere divides the chromosome unevenly into short (p, from *petite*) and long (q) arms which vary in length from chromosome to chromosome. Chromosomes with their centromere near the very end will often have satellites, small pieces of DNA attached to the rest of the chromosome by a stalk of multiple copies of ribosomal RNA.

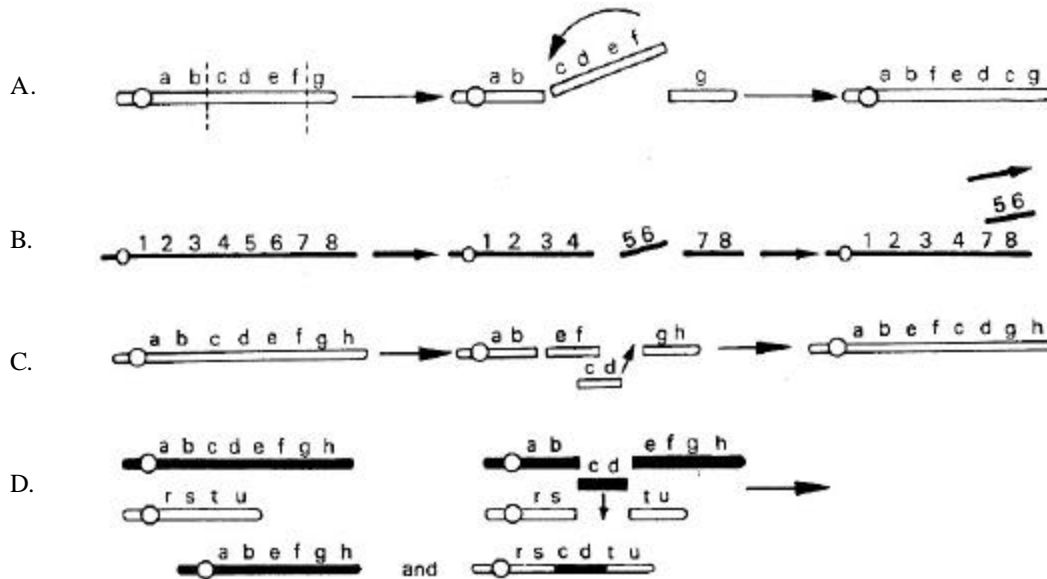
A standardized notation has been adopted to describe the banding pattern produced by Giemsa staining. In humans there are 23 pairs of chromosomes. Each autosome is assigned a number, 1 – 22 and the sex chromosomes are referred to as either X or Y. The arm of each chromosome is denoted with a “p” or “q,” thus, 7q would refer to the long arm of chromosome 7. Each arm is further divided into 1 to 3 regions, depending on the size. Numbering begins at the centromere and moves out toward the telomere (end). In this manner, 7q1 would refer to the first region on the long arm of chromosome 7. Each region is further divided into light and dark bands which are also numbered from proximal to distal (*e.g.* 7q11). Each band may be even further subdivided into subbands, which are denoted after a decimal point (7q11.2). As resolution of this technique increased, more numbers were added to represent even smaller divisions of the subbands (7q11.22) (Figure 2).

Figure 2 Designation of G bands on human chromosome 7. The constriction indicates the location of the centromere. Notation of the banding pattern is described in the text. Arrow represents the subband division 7q11.22.



During the course of meiosis, portions of chromosomes are often relocated, moving within the chromosome itself or between different chromosomes. This process produces changes in the morphology of the chromosome itself, which are referred to as chromosomal aberrations. There are a number of different types of modifications possible. In an **inversion**, a chromosome breaks at two places and the resulting small piece “flips over” in the other direction and is re-annealed within the chromosome (Figure 3). **Deletions**, as the name implies, involves the removal of a portion of chromosome that is subsequently lost. **Insertions**, on the other hand, are parts of unknown origin that become a part of an intact chromosome. In a **translocation**, a portion of chromosomal material is moved to a new location. It may be moved within the same chromosome or to a different one (Figure 3). If a mutual exchange occurs between two nonhomologous chromosomes the shift is termed a reciprocal translocation.

Figure 3 Formation of chromosomal aberrations. **A.** Inversion. **B.** Deletion. **C.** Translocation within the same chromosome. **D.** Translocation between chromosomes.



The banding patterns of chromosomes of humans, chimpanzees, gorillas and orangutans show a striking similarity which illustrates the close phylogenetic relationship of these species. Figure 4 contains a schematic representation of the chromosomes from these primates. They are arranged from left to right, as follows: human, chimpanzee, gorilla, and orangutan. The numbering (1-22, X, Y) reflects the system used to describe human chromosomes. Use Figure 4 to answer the questions that follow.

Questions

1. In which human chromosomes are satellites evident?
2. Which 3 chromosomes appear most similar in all 4 species?
3. How many pairs of chromosomes do normal humans have? How many are in the other 3 species? How can you tell?
4. Look at chromosome 1. How is the human chromosome different from the chimpanzee's? What is this aberration called? Make a red circle around the part of the chromosome with this aberration.

5. How does orangutan chromosome 3 differ from the others? What is this aberration called? Make a green circle around the aberration in the orangutan chromosome.

6. Use more green circles to indicate similar aberrations in chromosomes 4, 7, 8, 9, 12, and 18. These may be on chromosome of any of the 4 species.

7. Look closely at gorilla chromosomes 5 and 17. What has happened here? Use blue circles to indicate these aberrations.

8. Which chromosome and species shows a significant deletion on the q-arm? Indicate the deletion using an orange circle.

9. The difference in the appearance of human chromosome 2 and the other species can be explained by telomeric fusion. Based upon the appearance of the chromosomes, explain what happens during this process.

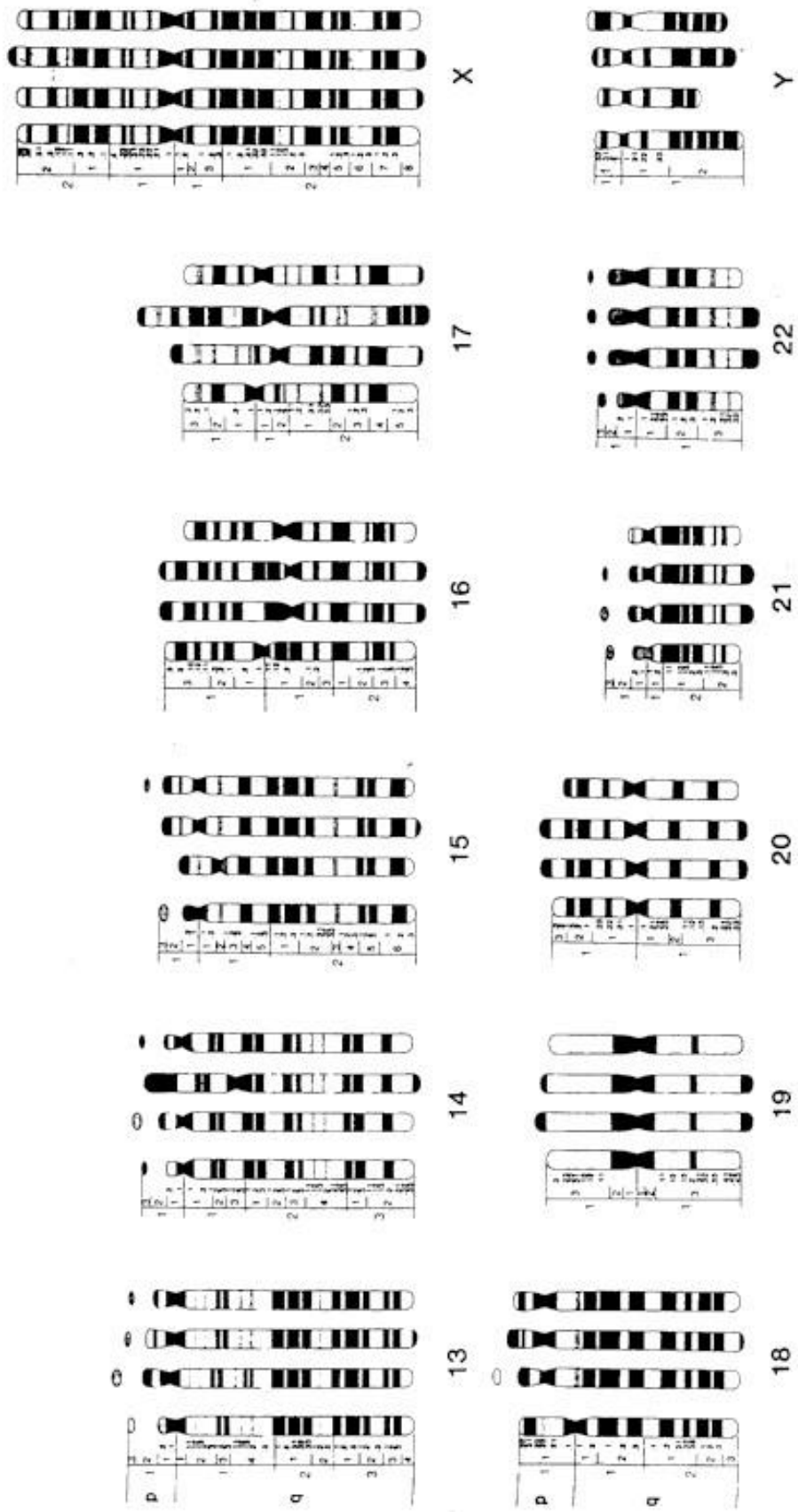
10. What does chromosome 2 indicate about the chromosomes of the common ancestors of humans, chimpanzees, gorillas and orangutans? What does it indicate about which species evolved most recently?

11. Fill in the squares in the key with the colors that indicate the type of aberration.

12. Look at orangutan chromosome 11. Describe what has happened here. Use your colors to indicate these changes.

Key

- Inversion
- Insertion
- Deletion
- Translocation



Key

