Autosomal Inheritance

Principles of Inheritance in Sexual Organisms (including humans)

An ancient sexual cycle governs the transmission of genetic information from generation to generation in humans and most other organisms excluding bacteria. Although there are lots of variations on the common theme, the general principles of inheritance in sexual organisms apply to single-celled fungi, plants, and animals including humans. At conception, an individual inherits from the mother a set of chromosomes present in the egg and from the father a matching (or homologous) set of chromosomes in the fertilizing sperm. DNA is the recipe for cellular and organismal function and a unique DNA molecule forms the centerpiece of each chromosome in each maternal and paternal set. Humans have 23 pairs of chromosomes with each chromosome pair having a distinct composition of functional gene regions. An individual inherits one member of each chromosome pair from their mother and the other member from their father. Considering each chromosome pair, only a single chromosome is subsequently transmitted to offspring. Meiosis is the specialized cell division responsible for sorting the 23 chromosome pairs so that each gamete (egg or sperm) contains one member of each pair. Fertilization brings these sets of chromosome together into pairs that are present inside all cells of an individual.

Essentially the same functional information is shared between the two members of 22 chromosome pairs. These are autosomes. Although the information encoded in the DNA base sequence for a gene present on a chromosome inherited from mom may differ from the DNA base sequence of the gene on the matching chromosome from dad, each parent contributes a copy. Furthermore, only one copy will be transmitted to a child regardless of the sex of that child. In contrast, the remaining pair of sex chromosomes contains a region that functions to determine the sex of an individual. The pair of sex chromosome in human males is mismatched, exhibiting a visible size difference and having almost entirely different DNA base sequences, and is designated as the X chromosome and the Y chromosome. Autosomes, the X chromosome, the Y chromosome, and the mitochondrial DNA molecule each exhibit distinct patterns of inheritance over multiple generations. However, autosomes comprise the vast majority of the genome and contain most of the sites examined in SNP tests offered by 23andMe, AncestryDNA, and the Family Finder test of Family Tree DNA.

Principles of Autosomal Inheritance

Two basic rules describe the inheritance of chromosomes. These are known as Mendel's Laws or Mendelian Principles named for their discoverer, Gregor Mendel, who performed and studied crosses in the garden pea. Discoveries in the early 20th century revealed the relationship between the rules of inheritance of traits described in 1866 by Mendel and the transmission of whole chromosomes. In the case of each chromosome pair, the modern principle of Segregation recognizes that only one member of the pair is transmitted to each gamete and that each member of the pair has an equal chance of being transmitted. The modern principle of Independent Assortment applies to the different pairs of chromosomes and considers that each pair of chromosomes segregates independently. As illustrated in the diagram below, the consequences of these principles can be seen when viewing inheritance over multiple generations. For each chromosome pair, only a single member is inherited from each parent and only a single member is transmitted to offspring. The chromosome sets inherited from parents undergo independent assortment that produces a combination of parental chromosomes transmitted to offspring.
Exchange between chromosome pairs also occurs to produce new combinations between the DNA inherited from each parent. During the specialized meiotic cell division, pairs of chromosomes associate with each other. Breaking of the parental DNA molecules and exchange between them produces a chromosome that retains all the functional information of each chromosome pair but in new combinations. In addition to the recombination due to independent assortment of different chromosome pairs, the recombination within chromosome pairs produces novel genetic combinations in each offspring. These processes of inheritance can be wonderfully visualized in direct comparisons of SNP data between related individuals.
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Recombination
Independent assortment and exchange between chromosome pairs forms new genetic combinations
Segregation of chromosomes ensures that you receive half of your autosomes from one parent and half from the other. However, segregation also results in the composition of your autosomes reflecting a diminishing contribution from each successive generation of your ancestors. A contribution of 25% of your autosomal regions is expected from each grandparent, 12.5% from each great grandparent, etc. Furthermore, beyond the strict 50% inheritance from parents, independent assortment and exchange between chromosomes pairs generates a large amount of uncertainty in the exact proportion of the autosomes contributed by each grandparent, great grandparent, etc. Thus, a large amount of variability is realized in the actual contribution from each ancestor. Some may have contributed a greater proportion than expected and some less, or in the case of distant ancestors, possibly no regions were inherited at all.
The Morgan as the Unit of Inheritance

The Morgan is a measure of chromosome length based on the amount of exchange between the chromosome pair during meiosis, so that 1 Morgan equals the distance between positions on a chromosome in which one point of recombination is expected per meiosis. This is an extremely useful measure for predicting the length of chromosome segments inherited from ancestors. On average, human autosomes are 1.6 Morgans and overall the 22 autosomal pairs contain 35.9 Morgans (Matise et al. 2007). During each meiosis, the human genome effectively segregates as 58 (i.e., 22 + 35.9) independent segments, each about 0.62 Morgans long.

Using the metric system for conversion, 1 Morgan equals 100 centiMorgans (cM). The average size of a chromosome segment transmitted through a single human meiosis is, therefore, 62 cM. Lengths of chromosomes expressed in Morgans or cM are obtained experimentally from measures of recombination between markers positioned along the chromosomes. Markers close to each other on the same chromosome and that experience only one recombination event per 100 meioses, i.e., 1% recombination, are approximately 1 cM separated. Take care applying this relationship between cM and recombination! Markers that are 100 cM apart do not exhibit 100% recombination, because the maximum recombination fraction between any two markers in the genome, including markers on separate chromosome pairs, is 50%. A mapping function models the relationship between recombination fraction and map distance.

The number of basepairs present in the DNA of a chromosome region measuring 1 cM is not uniform across the autosomes. Some regions are more refractory to recombination and have more basepairs per cM. Excluding these regions, the average relationship is 1.3 cM / Mbp (i.e., 1.3 cM / 1,000,000 basepairs). These two measures of human genome length, cM reflecting the frequency of exchange during meiosis and Mbp reflecting physical distance along the DNA, are commonly used in reference to the human genome including inferences from personal genome comparisons.
X Chromosome Inheritance

The X chromosome has a distinct pattern of inheritance. In the image below, the ancestral origins of the sex chromosomes are illustrated for a brother and sister. For the brother, his single X chromosome has its origins entirely from his maternal ancestry. Because the Y chromosome is transmitted from a father to a son, an X chromosome never passes through two consecutive generations of male ancestors. In contrast, the sister inherits an X from her father along with the X representing her maternal ancestry. Because females have a pair of X chromosomes, exchange can occur between the paternally and maternally inherited X chromosome through the same process that makes new combinations within autosomes.