Faculty Name: Dr. Kumari Sushma Saroj

Department: Zoology

College: Dr. L. K. V. D College, Tajpur, Samastipur

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Topic: Chromosome mapping

Chromosome mapping

Chromosome mapping is a technique used in autosomal DNA testing which allows the testee to determine which segments of DNA came from which ancestor.

Chromosome mapping is the assignment of genes to specific locations on a chromosome. A gene map serves many important functions and is much like understanding the basic human anatomy to allow doctors to diagnose patients with disease.

In order to map DNA segments on specific chromosomes it is necessary to test a number of close family relatives.

Ideally one should test both parents, one of their children, and a number of first to third cousins on both the maternal and paternal sides of the family.

Not everyone has close relatives available for testing or has the funds to pay for such testing. Indeed, even if you did test all of your first and second cousins you might not be able to map your entire genome.

In any case, the more first and second cousins you test, the higher the percentage of your genome that you can map, at least back to which parent or grandparent contributed any particular DNA segment.

Caution should be exercised when attempting to map smaller segments, and particularly segments under 10 cMs. You need to keep in mind that the shared segments as reported by the DNA testing companies are simply half-identical regions.

Half-identical regions (HIRs) may be either identical by descent (IBD) (true matches) or identical by state (IBS) (false positive matches).

Using phased data for the comparisons makes it much easier to determine which half-identical regions are identical by descent and identical by state.

Methodology

In order to map DNA segments it is necessary to maintain a list of your matches, preferably in a spreadsheet.

The matches spreadsheet should include the names and contact details of your matches, details of the matching segments and number of shared SNPs, the name of the most recent common ancestor and the known relationship.

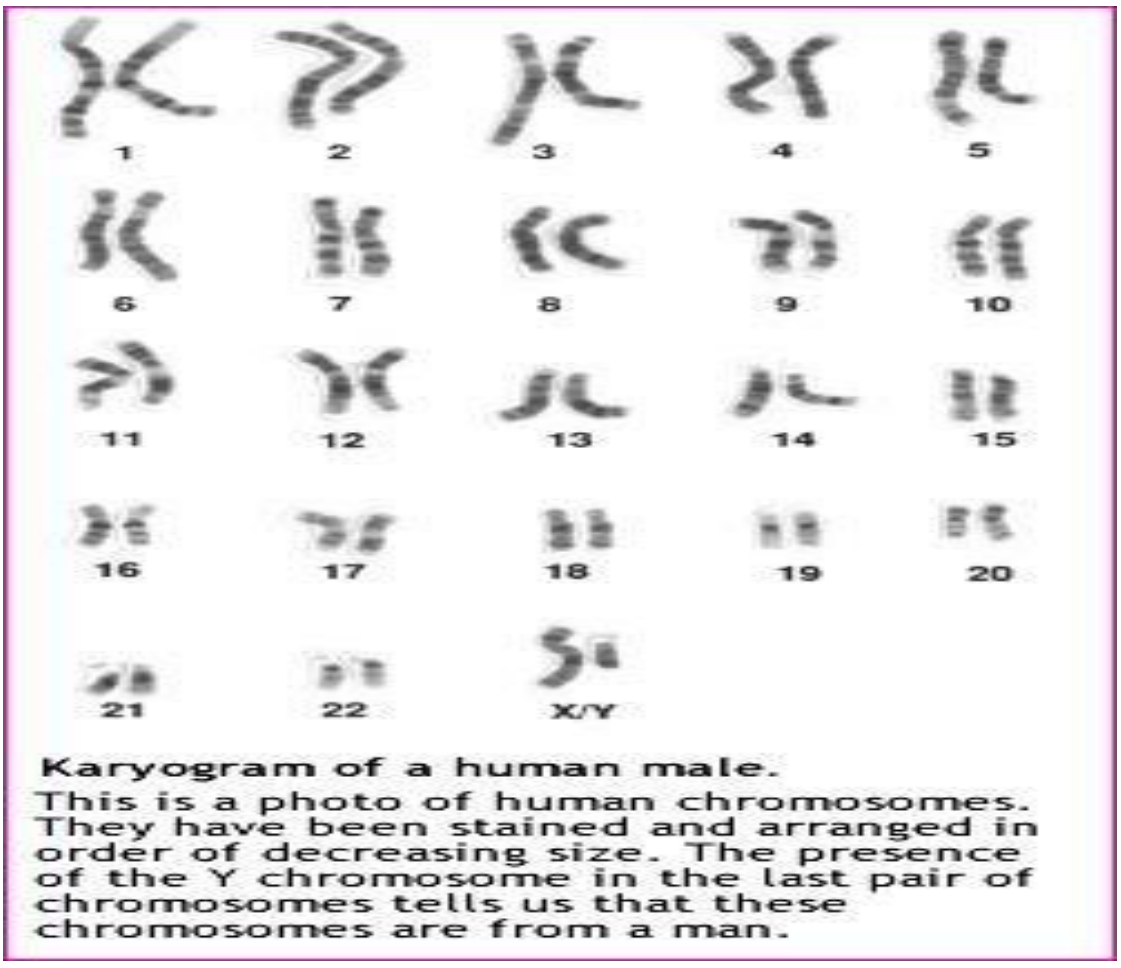
ISOGG member Tim Janzen has been one of the leading researchers using the technique of chromosome mapping.

Tim Janzen and Emily Aulicino have prepared a 16-page tutorial on the "Basics of chromosome mapping" which describes how to create a list of all one's matches and their accompanying HIR (half-identical regions) data.

Tim Janzen presented a lecture on chromosome mapping at Rootstock 2014 entitled "Advanced Techniques for Use of Autosomal DNA Tests to Break through Genealogical Brick Walls".

Visual phasing

Visual phasing is a methodology for assigning segments to specific grandparents based on the crossover points of three siblings. This technique can be employed when parents are not available for testing.



Karyogram of a human male.
This is a photo of human chromosomes. They have been stained and arranged in order of decreasing size. The presence of the Y chromosome in the last pair of chromosomes tells us that these chromosomes are from a man.

Gene mapping can provide clinicians with useful information regarding genes that are linked, or segregate closely together. Scientists use several methods to map genes to the appropriate locations.

These methods include family studies, somatic cell genetic methods, cytogenetic techniques, and gene dosage studies.

Family studies are used to determine whether two different genes are linked close together on a chromosome. If these genes are linked, it means they are close together on the same chromosome.

Additionally, the frequency with which the genes are linked is determined by recombination events (crossing over of the chromosomes during meiosis) between known locations or markers, and determines the linear order or genetic distance.

In somatic cell genetic methods, chromosomes are lost from a special type of cell and the remaining chromosome that has one gene, but not a different gene, would suggest that they are located on different chromosomes.

This method allows scientists to identify which chromosome contains the gene, and represents one of the first mapping methods used by scientists.

Cytogenetic techniques refer to utilization of karyotype preparations, a technique that allows scientists to visualize of chromosomes, using fluorescence so that a fluorescently-labeled gene will reveal where the gene is found on the chromosome.

Gene dosage studies use, for example, numerical abnormalities to determine indirectly the location of the gene on a chromosome. In Down syndrome, there can be three chromosome number 21 (Trisomy 21), resulting in three copies of the gene and therefore, three times as much protein.

In this case, a gene can be localized to chromosome 21 if there is three times as much protein in a cell with three 21 chromosomes. In this method, the amount of deoxyribonucleic acid (DNA) is assumed to be directly proportional to the amount of protein.

A cytogenetic map using these methods, various maps of chromosomes can be developed. These maps are called cytogenetic maps, linkage maps, physical maps, or a DNA sequence map.

A cytogenic map uses bands produced by a dye that stains chromosomes in a karyotpe and assigns genes to these bands.

A linkage map also referred to as a genetic map, orders genes along the DNA strand based on recombination frequency. Linkage mapping involves using two characteristics (and hence their responsible genes), both of which are present in one parent, combined with the frequency in which they occur together in the offspring to construct the map.

For example, the Moravian-born Augustinian monk and science teacher Gregor Johann Mendel (1823–1884) studied the flower color and plant height of peas.

He found that various heights were observed just as frequently with white flowers as with other colored flowers and similarly, dwarf plants occurred just as frequently with the two flower types.

Mendel concluded that the forms of the two genes were transmitted from parent to offspring independently of each other. This later became known as the Law of Independent Assortment, a concept that enhanced chromosome mapping techniques.

A physical map orders genes or markers along the DNA strand of a chromosome.

Finally, a DNA sequence, strung together, is the most precise type of map in that it contains both coding (gene-containing) and noncoding DNA.

It is felt that obtaining the complete DNA sequence from the genome of many different organisms will provide scientists with vital information that will unlock many biological mysteries.