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FLASH PRESENTATION NEW TEXT  
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WORD COUNT: APPROX 1700

NOTES:

- MENU—CHANGE TO:

#### GENETICS

Genes and Chromosomes

Dominant Transmission

Recessive Transmission

#### HSP AND THE NERVOUS SYSTEM

Nerve Signals to Muscles

(PLEASE DELETE THE “POPULATION” AND “FIRST SIGNS OF” PAGES; THIS INFORMATION IS BETTER COVERED ELSEWHERE ON THE SITE)

- With the exception of the pages to delete above, the pages and their order in this document correspond with the pages in the presentation. There is one page to be relocated, but it's treated according to where it appears now.
- On the “cast of characters” page (please change “cast of characters” to “key to demonstrations”), please arrange the graphics for the “cast” thus:  
Blue half-square, Pink half-square: normal gene (in son)  
Blue striped half-square, Pink striped half-square: mutated gene (in son)  
Blue half-circle, Pink half-circle: normal gene (in daughter)  
Blue striped half-circle, Pink striped half-circle: mutated gene (in daughter)
- On all other pages the graphics should appear basically the same.
- Please treat all text as “new” and completely replacing the text and notes that previously appeared on a page. Some of the pages with animation demonstrating possibilities for gene inheritance have new text that is either significantly longer or shorter than the original version. If there is more text than can fit attractively on a page, let me know and I'll cut it.
- Make sure to label all right arrows “Continue” so it's clear how to proceed to the next page in the presentation.

### **(GENES AND CHROMOSOMES PAGE 1)**

Humans have about 100,000 genes. These genes are the “blueprint” for our growth and physiological traits.

A chromosome is like a strand of genes. Genes come in pairs: Humans have 46 chromosomes arranged in 23 pairs in every cell in the body, except for the sperm and egg cells.

Sperm and egg cells contain just one of the chromosomes from each pair, i.e. 23 individual chromosomes. When a sperm and egg come together and conception begins, a new cell is formed that brings the individual chromosomes together into pairs. Thus, every person receives half of their genes from their father (from the sperm cell), and half from their mother (from the egg cell).

A genetic condition like HSP is caused by a mutated (changed) gene occurring on one or both of the chromosomes in a pair. If the mutated gene need only be present on one chromosome for the condition to exist, the condition is said to be *dominant* (strong). If the gene must be present on both chromosomes, it is said to be *recessive* (weak). Someone who has just one recessive HSP gene in a pair will not have HSP but will be a *carrier*. Since they usually show no signs of the condition, carriers may be unaware of what they carry. Lastly, if the mutated gene occurs on the X sex chromosome, effects vary by gender, and the condition is said to be “X-linked.” Most forms of HSP are dominant.

### **(CAPTION)**

One of the 22 homologous (similar paired) chromosomes or “autosomes.” (The X and Y sex chromosomes make up the 23<sup>rd</sup> pair).

**(GENES AND CHROMOSOMES PAGE 2: “DOMINANT CONDITIONS”)**

One of the genes for HSP that has been discovered exists on chromosome 2. The picture at right depicts a narrow red band on one allele (one half of the chromosome pair) that represents spastin, a dominant mutated gene for HSP. Since a dominant condition can be caused by just one gene in a pair, the owner of this chromosome would have HSP.

Dominant conditions tend to surface regularly in families. Recessive conditions sometimes arise unexpected, because genes can combine to produce a condition in a child when neither parent has the condition if both carry one recessive gene for it.

Below left is a graphic of chromosome 2, which illustrates how the pair is joined at the centromere (center point).

The graphic at below right is a simplified representation of the affected allele above, with the short arm and long arm labeled according to practice, listing the charted location of the spastin gene.

Total genes discovered on chromosome 2: 2,538 as of Sept. 26, 2002.

**(GENES AND CHROMOSOMES PAGE 3: “CAST OF CHARACTERS” [PLEASE CHANGE TO “KEY TO DEMONSTRATIONS”])**

The following demonstrations and charts will help you understand how genes for dominant and recessive conditions may be transmitted (passed down) in families. X-linked conditions will not receive coverage here.

Males will be represented by squares, and females by circles. Each shape also represents a pair of genes (in a pair of chromosomes) that may cause a condition such as HSP if one or both are mutated. As each egg or sperm contains just one or the other of a parent's set of genes, there are four possible gene combinations in a child. At each and every conception any one of these four outcomes again becomes a possibility, just as each child could be a boy or a girl, in a “random” event.

In the demonstrations each child will have one blue gene (from the father) and one pink gene (from the mother). Stripes denote a mutated gene, whether dominant or recessive. Children may be either “healthy” (have no mutated gene), “affected” (have one dominant mutated gene OR two recessive mutated genes) or “carriers” (have one recessive mutated gene).

The first demonstration will show possibilities for transmission in a family wherein one parent has one dominant mutated gene and therefore the condition it causes.

**DOMINANT TRANSMISSION PAGE 1: “DOMINANT TRANSMISSION”**

(PLEASE CHANGE “UNAFFECTED” LABEL TO “HEALTHY” AND ADD THE FOLLOWING NOTE)

1 The term “healthy” in these demonstrations simply means lacking any mutated gene for the form of the condition discussed.

### **(DOMINANT TRANSMISSION PAGE 2)**

Dominant forms of HSP are nearly always the result of just one gene. Chances are two out of four, or 50 percent, that each child born to a parent with dominant HSP will obtain the HSP gene and have the condition. There is the same chance that each child will inherit the affected parent's healthy gene and not have HSP.

Problems from dominant forms of HSP are usually fairly well known in a family, because the condition will not have skipped a generation in the past. On the other hand, if a person is seemingly the first in their family to have HSP, there is a possibility they have a new dominant gene mutation, but more likely their condition is recessive.

Recessive transmission will first be demonstrated in the case of one parent being a carrier (in HSP, having one recessive HSP gene).