Introduction to Mutations:

Deviations from the expected chromosomal number, or mutations in the structure of the chromosome, are inherited in predictable Mendelian fashion; they often result in dead organisms or substantial changes in phenotype.

Aneuploidy is the gain or loss of one or more chromosomes from the diploid amount, resulting in conditions of monosomy, trisomy, tetrasomy. Studies of monosomy (Turner Syndrome XO) and trisomic disorders (Down's Syndrome) have increased our understanding of the delicate balance that must exist in order for normal development to occur.

When complete sets of chromosomes are added to the diploid number, polyploidy is created. These sets may have identical or diverse genetic origin. Large segments of the chromosome may be modified by deletions or duplications.

Deletions may produce serious conditions such as Cri-du-chat Syndrome in humans.

Duplications may be important as a source of redundant or unique genes, but this usually has no effect on health. Inversions and translocations, while altering the gene order along chromosomes, cause no net loss of genetic information.

In an inversion, a sequence of genes is turned around. This does not affect health unless a critical gene sequence is physically disrupted.

Most children with Chronic Myeloid Leukemia have a translocation or mixed up chromosome, in which the tip of chromosome 22 is attached to chromosome 9. However, this type of heterozygous combination may cause genetically abnormal gametes during meiosis, often being lethal.

A change in chromosome number or in the arrangement of a chromosome region often results in phenotypic variation or disruption of development of an organism. Such phenotypic variations are passed to offspring in a predictable manner, resulting in many interesting genetic situations.

DNA structure and function
How DNA base sequence encodes information
Steps in DNA replication
Transcription - building an RNA chain
The genetic code - characteristics
Translation - from genetic message to protein product
Types of mutations - point mutations, altering number of DNA bases
Abnormal chromosome numbers: Aneuploidy, polyploidy
Deletions and duplications resulting from chromosome rearrangement - translocations, inversions

Gene Action/Mutations Worksheet:

Part 1: Answer the following questions.

1. Define a mutation.

Part 2: Gene Mutations

Note: Question #2 refers to the DNA master strand: AATGCCAGTGGTTCGCAC

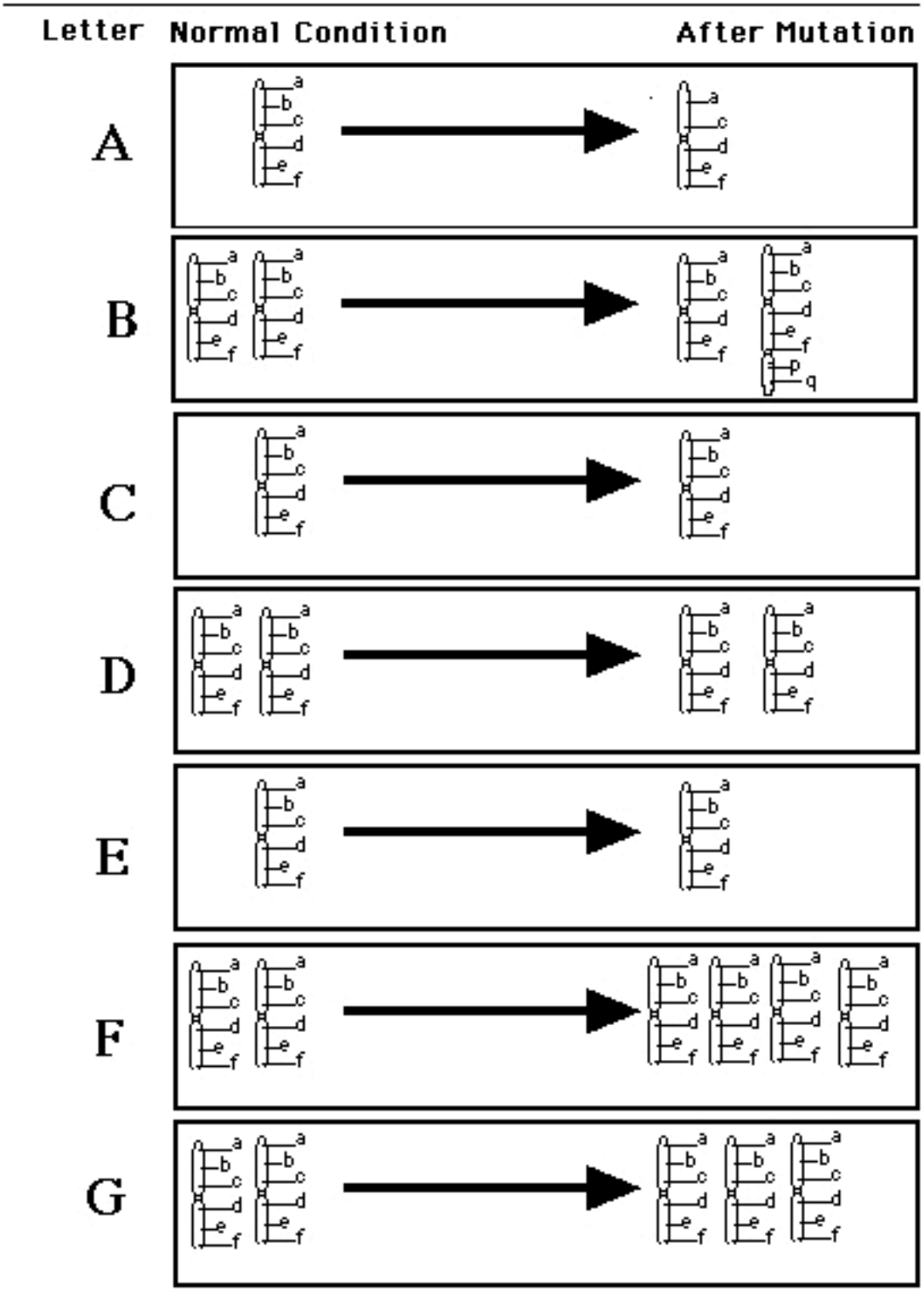
2. Write the nitrogen base sequence of the complementary DNA strand.

Question: #3 refer to the DNA master strand listed: AATGCCAGTGGTTCGCAC

3. If a `G' were added to the original master strand of DNA after the third nucleotide (T), what would the resulting mutated DNA strand look like? What kind of mutation is this?

4. If the `G' in the fourth nucleotide position were to be cut out of the original DNA strand, what would the resulting DNA strand look like? What kind of mutation is this?

Part 3: Chromosome Rearrangements



LETTER CHROMOSOME MUTATION DEFINITION SECTION:

5. Inversion - _____ 6. Polyploidy - _____ 7. Translocation - _____ 8. Trisomy -
9. Deletion - _____ 10. Monosomy - _____ 11. Nondisjunction -