Genetics

A. The Vocabulary of Genetics

1. Chromosome – bar-like structures of tightly coiled chromatin (DNA), visible during

cellular division.

2. Homologous chromosomes – chromosomes that carry the same genes and determine

the same traits. In a human, there are 23 homologous chromosome pairs for a total of

46 chromosomes.

A) Autosomes – of the 23 pairs of chromosomes present in human cells, 22 pairs of

them have nothing to do with the sex of the individual.

B) Sex chromosomes – of the 23 pairs of chromosomes present in human cells, 1 pair

determines the sex of the individual.

3. Gene – one of the biological units of heredity that determines the traits of an individual.

4. Alleles – different versions or alternate forms of a gene (example: blue eyes, brown

eyes, green eyes, etc). Alleles are often designated as being dominant or recessive.

A) Dominant alleles can mask the effects of other alleles (example: widow’s peak).

These alleles are always designated with a capital letter such as “A”.

B) Recessive alleles are easily masked by the effects of other alleles (example: straight

hairline). These alleles are always designated with a lower case letter such as “a”.

5. Genotype – a person’s combination of alleles.

A) Homozygous dominant – the person possesses two dominant alleles (AA).

B) Heterozygous – the person possesses one dominant allele and one recessive allele

(Aa).

C) Homozygous recessive – the person possesses two recessive alleles (aa).

6. Phenotype – a person’s physical expression of their genetic make-up (genotype).

A) A homozygous dominant person shows the dominant trait on the outside.

B) A homozygous recessive person shows the recessive trait on the outside.

C) A heterozygous person often shows only the dominant trait on the outside because

the dominant allele masks the presence of the recessive.

B. Types of Inheritance

1. Dominant-Recessive Inheritance

A) This type of inheritance is common with traits like hitchhiker thumb, rolling the

tongue, hairline, PTC tasting, etc.

B) In every case, the dominant trait masks the recessive.

C) Punnett Squares are useful tools for determining genetic inheritance of

dominant/recessive traits.

1) However, Punnett Squares only predict the probability of having a certain

percentage of offspring with a particular genotype or phenotype.

2) The larger the number of offspring, the more likely that the ratios will conform

to the predictions.

2. Incomplete Dominant Inheritance

A) Recessive traits are not masked in the heterozygous form. Instead, a third

phenotype (different from either the dominant or recessive one) is produced.

B) Example: A + B = C

C) Example: Red flowers + White flowers = Pink flowers

D) Sickle cell anemia is a human condition associated with this type of inheritance

where the intermediate form has sickle cell trait but not full blown sickle cell

disease.

3. Codominant Inheritance and Multiple Alleles

A) There are some traits that demonstrate more than two dominant alleles.

B) Blood type is a common multiple allele pattern of inheritance.

C) Because two alleles are “dominant” neither has the ability to mask the other;

instead they are codominant and mixture of the two alleles shows up in the

phenotype of the offspring.

D) Example: A + B = AB

E) Example: Red flower + White flower = Red flower with white spots.

4. Sex-linked Inheritance

A) All of the other patterns of inheritance mentioned above are demonstrations of

genes carried on autosomal chromosomes and an individual has equal chances of

getting the gene whether that person is male or female.

B) Sex-linked inheritance however, demonstrates traits that are carried on the sex

chromosomes and an individual’s chance of getting the trait varies with the sex of

the individual.

C) Most sex-linked traits are carried on the X chromosome while very few are carried

on the Y chromosome.

D) X-linked traits affect both males and females because both sexes will receive at

least one X in their genotype (XX=females; XY=males).

1) Ex: hemophilia and color blindness

E) Y-linked traits only affect males because females do not receive a Y chromosome.

1) Ex: hair on the ear lobes

5. Polygenic Inheritance

A) Polygenic inheritance is the result from several different gene pairs at different

locations within the genetic makeup work together to produce a particular

phenotype.

B) Ex. over 150 different genes influence eye, hair and skin colors.

C) Different combinations produce variations in skin color from dark skin to light

skin.

6. However, environment can affect the expression of genes.

A) Maternal drug use can alter normal gene expression during embryonic

development.

B) Nutrition and diet

C) Hormonal deficits and excesses

C. Sources of Genetic Variation

1. Mendel’s Law of Segregation

A) Each organism contains two factors (alleles) for each trait and these randomly align

along the metaphase plate.

B) The factors then segregate during the formation of gametes so that each gamete

contains only one factor for each trait.

C) This reshuffling of the factors helps explain how variations come about and why

offspring differ from their parents.

2. Law of Independent Assortment

A) Members of one pair of factors separate independently of members of another pair

of factors.

B) Therefore, all possible combinations of factors can occur in the gametes.

3. Crossing over of Homologous resulting in gene recombination

A) During Meiosis I (prophase I), two of the four chromatids (one maternal & the

other paternal) may cross over at one or more points and exchange corresponding

gene segments.

B) The recombinant chromosomes contain new gene combinations, adding to the

variability arising from independent assortment.

4. Random Fertilization

A) The third source of genetic variation is random fertilization of eggs by sperm.

B) When just considering independent assortment, crossing over and random

fertilization, any resulting offspring represents ONE out of the close to 72

TRILLION zygotes possible.