BIO 120: VARIATION

WHERE DOES IT COME FROM?
HOW IS IT INHERITED?
HOW DOES IS IT AFFECT TRAIT VARIABILITY?
AND MORE

REQUIREMENTS FOR DARWIN'S THEORY:

- VARIATION
- HEREDITY
- SELECTION

SOME BASIC TERMS USED IN GENETICS:

- **GENOTYPE**
  GENETIC CONSTITUTION
- **PHENOTYPE**
  THE ORGANISM AS OBSERVED
- **GENOME**
  ENTIRE ORGANISM'S DNA INCLUDING BOTH GENES AND NON-CODING REGIONS

GENE

1. THE FUNCTIONAL UNIT OF INHERITANCE
2. A DNA SEQUENCE COMPRISED OF CODONS ASSOCIATED FOR A SPECIFIC BIOLOGICAL FUNCTION.

VARIATION ⟶

- MUTATION
- RECOMBINATION
- GENE FLOW
- HYBRIDIZATION

IMPORTANCE DEPENDS ON THE TIME SCALE

INDEPENDENT ASSORTMENT & RECOMBINATIONS

n = 23 CHROMOSOMES, 2^{23} > 8 MILLION POSSIBE GAMETE COMBINATIONS

BARBARA McCLINTOCK

WON THE 1983 NOBEL PRIZE FOR PHYSIOLOGY – MEDICINE FOR

JUMPING GENES OR TRANSPOSABLE GEN. MUT. IN MAZE.

A MUTANT IN THE FIELD – WHAT WILL HAPPEN TO IT?

- IF POLLINATORS CONTINUE TO FEED BY, A MUTATION STAYS. → FIXATION? → persists
- IF NO POLLINATORS, THE MUTATION IS TAKEN OUT

MUTATION =>

1. STABLE CHANGE IN DNA SEQUENCE
2. OCCURS AT A LOW RATE
3. 4 POSSIBLE OUTCOMES.
   - NEUTRAL
   - DELETERIOUS
   - LETHAL
   - BENEFICIAL

4) TO BE IMPORTANT FOR EVOLUTION, MUST OCCUR IN GERM CELLS — SOMATIC MUTATIONS NOT INHERITED

→ MANY MUTATIONS ARE CONTEXT - DEPENDENT.

→ COPYING ERRORS IN DNA.

5) ENVIRONMENTAL CHANGES AFFECT THE MUTATION RATES.

- WHAT IS THE METHOD OF INHERITANCE?
- HOW ARE TRAITS EXPRESSED IN PARENTS AND OFFSPRING?

GREGOR MENDEL 1822-1884

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F1 = FIRST FILIAL GENERATION(S)

BLENDING INHERITANCE OCCURS WHEN MIXING OF A CROSS SHOWS INTERMEDIATE ATTRIBUTES

→ MENDEL'S RESULTS DO NOT CONFORM TO PREDICTIONS OF BLENDING INHERITANCE.

MENDEL'S CONCLUSIONS

1. THERE ARE 'GENES' WHICH DETERMINE INHERITANCE
2. MOST ORGANISMS CARRY 2 COPIES OF EACH GENE (ALLELES) AND ARE DIPLOID.
3. ORGANISMS PRODUCE GAMETES
4. OFFSPRING INHERIT ONE ALLELE FROM EACH PARENT AT RANDOM

MUTATION & STRUCTURE DNA

1. POINT MUTATION : AT GCA GT - ACCAGT

2. INSERTIONS / DELETIONS (INCLUDING JUMPING GENE) & MORE

MUTATION RATES IN EUKARYOTES

1,6 - 2.0 IN HUMANS PER POPULATION.

- EACH HUMAN CARRIES 3-5 RECESSIVE LETHAL ALLELS
- ALLELES CAUSING DEATH WHEN HOMOZYGOUS
- MATING AMONG RELATIVES CAUSES A HIGHER INCIDENCE OF OFFSPRING MORTALITY.

NEW ZEALAND

SPENCER WORKS ON INBREEDING

→ SABBATICAL IN HARVARD

→ STATES DETERMINE THE PERMISSIBILITY OF INBREEDING

→ THE DIFFERENCE IN MORTALITY RATES LINKED TO THE PORTION OF INBREEDING

DISCRETE VS CONTINUOUS TRAITS

DISCRETE TRAITS INHERITED BY 1 OR 2 GENES (MAJOR GENES)

CONTINUOUS TRAITS — COMPLEX INHERITANCE BY MANY GENES (POLYGENES) OF SMALL EFFECT — QUANTATIVE INHERITANCE

GENETIC POLYMORPHISM

INVOLVES DISCRETE PHENOTYPES GOVERNED BY SEGREGATION OF A SMALL NUMBER OF ALLELES AT 1-2 GENES.

EG

WILD GLADIOUS COLOURS OR HUMAN SEX.

65% OF VARIATION IN HUMAN HEIGHT IS HERITABLE — AN EXAMPLE OF QUANTITATIVE INHERITANCE.

GENE NUMBER & PHENOTYPIC DISTRIBUTION

RELATION BETWEEN NUMBER OF GENES CONTROLLING A TRAIT AND PHENOTYPIC VARIABILITY

DISCRETE

- MAJOR GENES,
- DOMINANCE AND RECESSIVENESS
- GENETIC POLYMORPHISM

QUANTATIVE

- POLYGENES
- SELECTION RESPONSE
- ARTIFICIAL SELECTION

SIR RONALD FISHER

'THE RATE OF INCREASE IN FITNESS OF A POPULATION AT ANY TIME IS EQUA TO THE MEASURE OF GENETIC VARIATION'