

Mendelian Genetics

- Genes - code for proteins
 - Each normal sperm and egg contains a recipe for building a living organism
 - NOT a "blueprint" as is often said
- Prokaryotes such as bacteria typically contain a single circular bacteria chromosome
- Eukaryotes contain multiple linear chromosomes
 - Chromosome numbers do not necessarily correlate to the genetic complexity
 - Humans have 46 chromosomes
 - A fern has 1260
- Gene mutation creates genetic diversity
 - Copying and transmitting genes from one generation to the next increases genetic variation
 - Mutations occur during copying and lead to different alleles
 - Can be beneficial, neutral, or dysfunctional
- Segregation and independent assortment create gamete diversity
 - Both parents make a contribution, but not 100% contribution
 - While they only contribute half of the genetic information, they give out a complete copy of the "recipe book"
 - 23 "pages" from mom and 23 "pages" from dad
 - During meiosis the separation of pairs of alleles is known as segregation
 - Why siblings are not identical (besides identical twins)
- independent assortment → genes are (mostly) inherited independently of each other
- Segregation dictates that alleles will have a 50% chance of ending up in a gamete (sperm or egg)
 - 23 chromosome pairs
 - = 8,000,000 possible unique gametes
 - Those gametes undergo random fertilization this means that gamete will fertilize no matter what alleles they contain
 - 1/64,000,000

The Inheritance of traits

- Gregor Mendel (1822)
 - Grew 30,000 pea plants and documented the inheritance of their traits after transplanting pollen
 - Had no idea about genes but his experiments were sound and rigorous
 - Genotype - the genetic composition of the organism (physical genes and their codes)
 - A, G, T, C
 - Phenotype - physical traits prescribes for those genes (can see with the naked eye)

Medelian Genetics Study Guide

NARAYAN CHANGDER



Medelian Genetics Study Guide:

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Why Do Genetics Matter to You This book is a summary of The Gene An Intimate History by Siddhartha Mukherjee Siddhartha Mukherjee s book chronicles the fascinating history of discovery in classical genetics molecular genetics genetic engineering and the human genome project It shows How our genes and the environment define our identities and personalities How genetic engineering technologies can be used to manufacture drugs safely and How genetic diagnosis and gene therapies can be used to treat complex genetic diseases Genetics is at the frontiers of science today and its impact is often misunderstood The public is often misled by science fiction and remains largely in the dark as to the actual

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Mendelian Inheritance in Man Victor A. McKusick, 1998-06-29 The twelfth edition of this classic reference work includes More than 2 000 new entries A total of more than 9 000 entries New features and enhancement of the familiar old features Mapping information on more than 4 000 genes of known function Information on specific point mutations responsible for more than 700 genetic disorders or neoplasms Mendelian Inheritance in Man MIM is a genetic knowledgebase that serves clinical medicine and biomedical research including the Human Genome Project It aims to be comprehensive not only complete but also collated integrated and interpreted authoritative not only accurate but also sound in its interpretations and judgements and timely not only up to date but also historically dimensioned From a review of the eleventh edition Reproductive Toxicology Even the convenience of computer based forms of MIM cannot eliminate the need for MIM in book form The preface provides a wonderful synopsis of human genetics The information contained in this text serves as a concise review for those with a genetics background From a review of the tenth edition New England Journal of Medicine Victor McKusick has been for all these years the shepherd of the development of the field of clinical genetics Perhaps his most important pragmatic achievement has been the 10 editions of Mendelian Inheritance in Man which rapidly became and has remained the principal source of information on inherited diseases for all clinical geneticists In addition to the erudite entries in the books the references given with each description represent a magnificent bibliography of clinical genetics With McKusick's leadership and continued interest in gene mapping the book also represents an important compendium of the location of genes on specific chromosomes The book is a magnificent security blanket for the clinical geneticist and should be in the libraries not only of these specialists but also of all others who see patients with diseases that have genetic components

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