### Agenda 12/1

- 1) Check project outlines while you share yours with neighbors I'll collect any that you want more detailed feedback on
- 2) Start Development Powerpoint –watch video and do slides

http://www.youtube.com/watch?v= 22CFCxDUy0

Note - during slides take notes when indicated to study for quiz

Homework -

- •Finish 18.4 and 47.3 notes, concept checks due tomorrow, online assignment due Wed
- Short Quiz Wed.
- Ch. 14 Notes and concept checks due Friday, Ch. 14 online assignment due Monday (long! 2 hours)

Development occurs at many points in the life cycle of an animal

- This includes metamorphosis and gamete production, as well as embryonic development
- •Although animals display different body plans, they share many basic mechanisms of development and use a common set of regulatory genes
- •Biologists use **model organisms** to study development, chosen for the ease with which they can be studied in the laboratory

### **Fertilization**

- Molecules and events at the egg surface play a crucial role in each step of fertilization
  - Sperm penetrate the protective layer around the egg
  - •Receptors on the egg surface bind to molecules on the sperm surface
  - Changes at the egg surface prevent polyspermy,
     the entry of multiple sperm nuclei into the egg

### Cleavage

- •Fertilization is followed by **cleavage**, a period of rapid cell division without growth
- Cleavage partitions the cytoplasm of one large cell into many smaller cells called blastomeres
- The solid ball of cells is called a morula
- The blastula is a ball of cells with a fluid-filled cavity called a blastocoel

## •A blastocoel forms within the morula → blastula Yolk least concentrated here

Polarity (common in animals beside mammals) determines planes of cleavage.

Yolk least concentrated here, also may become anterior side of animal

Yolk most concentrated here

- The three layers produced by gastrulation are called embryonic germ layers
  - The ectoderm forms the outer layer
  - •The endoderm lines the digestive tract
  - •The **mesoderm** partly fills the space between the endoderm and ectoderm
- Each germ layer contributes to specific structures in the adult animal

### Go to study area online – Ch. 47

Watch video of sea urchin development

### Organogenesis

- During organogenesis, various regions of the germ layers develop into rudimentary organs
- •Early in vertebrate organogenesis, the **notochord** forms from mesoderm, and the neural plate forms from ectoderm

## NOW – WATCH FROG DEVELOPMENT VIDEO FROM WEBSITE – ALSO FETAL ULTRASOUNDS

### c. Programmed cell death (apoptosis) plays a role in the normal development and differentiation.

Students should be able to demonstrate understanding of the above concept by using an illustrative example such as:

- Morphogenesis of fingers and toes
- Immune function
- C. elegans development
- Flower development

**X X** Names of the specific stages of embryonic development are beyond the scope of the course and the AP Exam.

### Programmed Cell Death

- Programmed cell death is also called apoptosis
- •At various times during development, individual cells, sets of cells, or whole tissues stop developing and are engulfed by neighboring cells
- •For example, many more neurons are produced in developing embryos than will be needed
  - -Extra neurons are removed by apoptosis
- •Another example is the morphogenesis of fingers and toes (cells between undergo apoptosis)

## Apoptosis (Cell suicide) and Normal Development

- •A built-in cell suicide mechanism is essential to development in all animals.
  - Similarities between the apoptosis genes in mammals and nematodes indicate that the basic mechanism evolved early in animal evolution.
  - The timely activation of apoptosis proteins in some cells functions during normal development and growth in both embryos and adults.
    - It is part of the normal development of the nervous system, normal operation of the immune system, and for normal morphogenesis of human hands and feet.

- Problems with the cell suicide mechanism may have health consequences, ranging from minor to serious.
  - Failure of normal cell death during morphogenesis of the hands and feet can result in webbed fingers and toes.
  - Researchers are also investigating the possibility that certain degenerative diseases of the nervous system result from inappropriate activation of the apoptosis genes.
  - Others are investigating the possibility that some cancers result from a failure of cell suicide which normally occurs if the cell has suffered irreparable damage, especially DNA damage.

### Agenda 12/2

- •Warm- up: Describe and name the major stages of embryological development starting with fertilization
- •Continue development through bicoid I check notes during this
- Practice questions
- •Start 47.3 slides aim to get through fate mapping

- •Homework -
- Online assignment due tomorrow
- •Study for small quiz tomorrow- 47.3 and 18.4
- Ch. 14 Notes and concept checks due Friday, Ch. 14 online assignment due Monday (long! 2hrs.)

# Concept 18.4: A program of differential gene expression leads to the different cell types in a multicellular organism

- During embryonic development, a fertilized egg gives rise to many different cell types
- •Cell types are organized successively into tissues, organs, organ systems, and the whole organism
- Gene expression orchestrates the developmental programs of animals

### Genes and Development

Differential gene expression leads to different cell types in multicellular organisms

- •Zygote undergoes transformation through 3 interrelated cell processes
  - 1. Cell division mitosis increases # of cells
  - 2. **Morphogenesis** organization of cells into tissues and organs
  - 3. **Cell differentiation** cells become specialized in structure and function
    - What are the terms that describe an embryo going through these stages?

- •Cell differentiation is the process by which cells become specialized in structure and function
- •The physical processes that give an organism its shape constitute **morphogenesis**
- •Differential gene expression results from genes being regulated differently in each cell type
- Materials in the egg can set up gene regulation that is carried out as cells divide

## Cytoplasmic Determinants and Inductive Signals

TAKE NOTES

- An egg's cytoplasm contains RNA, proteins, and other substances that are distributed unevenly in the unfertilized egg
- •Cytoplasmic determinants are maternal substances in the egg that influence early development
- •As the zygote divides by mitosis, cells contain different cytoplasmic determinants, which lead to different gene expression

#### TAKE NOTES

- •The other important source of developmental information is the environment around the cell, especially signals from nearby embryonic cells
- •In the process called **induction**, signal molecules from embryonic cells cause transcriptional changes in nearby target cells
- •Thus, interactions between cells induce differentiation of specialized cell types

# Sequential Regulation of Gene Expression During Cellular Differentiation

- Determination commits a cell to its final fate
- Determination precedes differentiation
- •Cell differentiation is marked by the production of tissue-specific proteins

TAKE NOTES

# Pattern Formation: Setting Up the Body Plan

- Pattern formation is the development of a spatial organization of tissues and organs
- In animals, pattern formation begins with the establishment of the major axes
- •Positional information, the molecular cues that control pattern formation, tells a cell its location relative to the body axes and to neighboring cells

KNOW THESE VOCAB WORDS

-THEY SHOULD BE IN YOUR

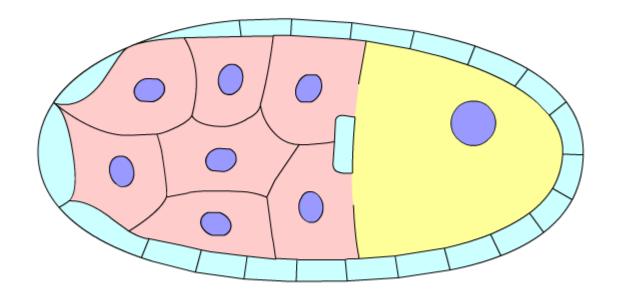
CORNELL NOTES

## GENETIC ANALYSIS OF EARLY DEVELOPMENT: SCIENTIFIC INQUIRY

- •Edward B. Lewis, Christiane Nüsslein-Volhard, and Eric Wieschaus won a Nobel Prize in 1995 for decoding pattern formation in *Drosophila*
- •Lewis discovered the **homeotic genes**, which control pattern formation in late embryo, larva, and adult stages
- They found 120 genes essential for normal segmentation

### Axis Establishment

- •Maternal effect genes encode for cytoplasmic determinants that initially establish the axes of the body of *Drosophila*
- •These maternal effect genes are also called **egg- polarity genes** because they control orientation of the egg and consequently the fly



Ch. 18 slide 106

Animation: Development of Head-Tail Axis in Fruit Flies Right-click slide / select "Play"

## Bicoid: A Morphogen Determining Head Structures

- One maternal effect gene, the bicoid gene, affects the front half of the body
- •An embryo whose mother has no functional *bicoid* gene lacks the front half of its body and has duplicate posterior structures at both ends

- •This phenotype suggests that the product of the mother's *bicoid* gene is concentrated at the future anterior end
- •This hypothesis is an example of the morphogen gradient hypothesis, in which gradients of substances called **morphogens** establish an embryo's axes and other features

TAKE NOTES – KNOW BICOID EXAMPLE

- •The bicoid research is important for three reasons
  - It identified a specific protein required for some early steps in pattern formation
  - It increased understanding of the mother's role in embryo development
  - It demonstrated a key developmental principle that a gradient of molecules can determine polarity and position in the embryo

PAUSE HERE FOR STUDY BREAK

Essential knowledge 2.E.1: Timing and coordination of specific events are necessary for the normal development of an organism, and these events are regulated by a variety of mechanisms.

a. Observable cell differentiation results from the expression of genes for tissue-specific proteins.

b. Induction of transcription factors during development results in sequential gene expression.

Evidence of student learning is a demonstrated understanding of each of the following:

- 1. Homeotic genes are involved in developmental patterns and sequences.
- 2. Embryonic induction in development results in the correct timing of events.
- 3. Temperature and the availability of water determine seed germination in most plants.
- 4. Genetic mutations can result in abnormal development.
- 5. Genetic transplantation experiments support the link between gene expression and normal development.
- 6. Genetic regulation by microRNAs plays an important role in the development of organisms and the control of cellular

## Learning Objectives

**LO 2.31** The student can connect concepts in and across domains to show that timing and coordination of specific events are necessary for normal development in an organism and that these events are regulated by multiple mechanisms. [See **SP 7.2**]

**LO 2.32** The student is able to use a graph or diagram to analyze situations or solve problems (quantitatively or qualitatively) that involve timing and coordination of events necessary for normal development in an organism. [See **SP 1.4**]

**LO 2.33** The student is able to justify scientific claims with scientific evidence to show that timing and coordination of several events are necessary for normal development in an organism and that these events are regulated by multiple mechanisms. [See **SP 6.1**]

**LO 2.34** The student is able to describe the role of programmed cell death in development and differentiation, the reuse of molecules, and the maintenance of dynamic homeostasis. [See **SP 7.1**]

## Essential knowledge 4.A.3: Interactions between external stimuli and regulated gene expression result in specialization of cells, tissues and organs.

- a.Differentiation in development is due to external and internal cues that trigger gene regulation by proteins that bind to DNA. [See also 3.B.1, 3. B.2]
- Structural and functional divergence of cells in development is due to expression of genes specific to a particular tissue or organ type. [See also 3.B.1, 3.B.2]
- c. Environmental stimuli can affect gene expression in a mature cell. [See also **3.B.1**, **3.B.2**]

### Learning Objective:

**LO 4.7** The student is able to refine representations to illustrate how interactions between external stimuli and gene expression result in specialization of cells, tissues and organs. [See **SP 1.3**]

### 12/3

- Study 5 min
- •Quiz (15 min)
- •Finish 47.3 slides (35 min) students explain pictures
- If finish, start Genetics practice

#### Homework –

Genetics practice packet pg. 1 due tomorrow

Ch. 14 Notes and concept checks due Friday, Ch. 14 online assignment due Monday (long! 2hrs)

Ch. 15 Notes, concept checks due next Thurs and online assignment due next Friday

•Next Unit Test (Ch. 11-15, 18.4, 47.3) approx. 12/16, be working on review manual chapters 11-15 (will be collected day of test)

# Concept 47.3: Cytoplasmic determinants and inductive signals contribute to cell fate specification

- •Determination is the term used to describe the process by which a cell or group of cells becomes committed to a particular fate
- Differentiation refers to the resulting specialization in structure and function

### Fate Mapping

- •Fate maps are diagrams showing organs and other structures that arise from each region of an embryo
- Classic studies using frogs indicated that cell lineage in germ layers is traceable to blastula cells

- •Later studies of *C. elegans* used the ablation (destruction) of single cells to determine the structures that normally arise from each cell
- •The researchers were able to determine the lineage of each of the 959 somatic cells in the worm

### Axis Formation

- A body plan with bilateral symmetry is found across a range of animals
- This body plan exhibits asymmetry across the dorsal-ventral and anterior-posterior axes
- The right-left axis is largely symmetrical

Yesterday, we talked about this in the fruit fly with the bicoid example. Today we look at axis formation in vertebrates.

#### Skip to picture

- •The anterior-posterior axis of the frog embryo is determined during oogenesis
- •The animal-vegetal asymmetry indicates where the anterior-posterior axis forms (animal is embryo, vegetal is yolk)
- The dorsal-ventral axis is not determined until fertilization

- Upon fusion of the egg and sperm, the egg surface rotates with respect to the inner cytoplasm
- This cortical rotation brings molecules from one area of the inner cytoplasm of the animal hemisphere to interact with molecules in the vegetal cortex
- This leads to expression of dorsal- and ventralspecific gene expression

#### OTHER VERTEBRATES

- In chicks, gravity is involved in establishing the anterior-posterior axis
- Later, pH differences between the two sides of the blastoderm establish the dorsal-ventral axis
- •In mammals, experiments suggest that orientation of the egg and sperm nuclei before fusion may help establish embryonic axes

## Restricting Developmental Potential

- •Hans Spemann performed experiments to determine a cell's developmental potential (range of structures to which it can give rise)
- Embryonic fates are affected by distribution of determinants and the pattern of cleavage
- The first two blastomeres of the frog embryo are totipotent (can develop into all the possible cell types)

- In mammals, embryonic cells remain totipotent until the 8-cell stage, much longer than other organisms
- Progressive restriction of developmental potential is a general feature of development in all animals
- In general tissue-specific fates of cells are fixed by the late gastrula stage

# Cell Fate Determination and Pattern Formation by Inductive Signals

 As embryonic cells acquire distinct fates, they influence each other's fates by induction

## The "Organizer" of Spemann and Mangold

- •Spemann and Mangold transplanted tissues between early gastrulas and found that the transplanted dorsal lip triggered a second gastrulation in the host
- •The dorsal lip functions as an organizer of the embryo body plan, inducing changes in surrounding tissues to form notochord, neural tube, and so on

## Formation of the Vertebrate Limb

- Inductive signals play a major role in pattern formation, development of spatial organization
- •The molecular cues that control pattern formation are called **positional information**
- •This information tells a cell where it is with respect to the body axes
- •It determines how the cell and its descendents respond to future molecular signals

- The wings and legs of chicks, like all vertebrate limbs, begin as bumps of tissue called limb buds
- The embryonic cells in a limb bud respond to positional information indicating location along three axes
  - Proximal-distal axis
  - Anterior-posterior axis
  - Dorsal-ventral axis

- One limb bud-regulating region is the apical ectodermal ridge (AER)
- The AER is thickened ectoderm at the bud's tip
- The second region is the zone of polarizing activity (ZPA)
- •The ZPA is mesodermal tissue under the ectoderm where the posterior side of the bud is attached to the body

- •Tissue transplantation experiments support the hypothesis that the ZPA produces an inductive signal that conveys positional information indicating "posterior"
- Sonic hedgehog is an inductive signal for anterior/posterior limb development – gradient from ZPA
- Hox genes also play roles during limb pattern formation

## Cilia and Cell Fate

- Ciliary function is essential for proper specification of cell fate in the human embryo
- Motile cilia play roles in left-right specification (always sweep fluid to the left, creating slight asymmetry of morphogens on left-right axis)
- Monocilia (nonmotile cilia) are on every cell and act as antenna to receive signal proteins (such as Sonic hedgehog)
  - play roles in normal kidney development

## Agenda 12/4

- •Gregor Mendel online cd activity 4-2-2 (15 minutes) keep a list of main ideas
- •Mendel slides (skim Ch. 14 Pwpt. Slides 19-35) (10 min)
- •Genetics Practice Problems Key questions page 1? (10 min)
- •Do Dihybrid cross together using rule of multiplication/addition (5min)
- Modes of Inheritance/Pedigrees (slides follow) (15 min)
- •X-Linked color-blind problem (Sex-Linked Review) last 5 min

Homework –

Punnett square practice pg. 2 due tomorrow

- Ch. 14 Notes and concept checks due tomorrow, Ch. 14 online assignment due Monday (long! 2hrs)
- Ch. 15 Notes, concept checks due next Thurs and online assignment due next Friday
- •Next Unit Test (Ch. 11-15, 18.4, 47.3) approx. 12/16, be working on review manual chapters 11-15 (will be collected day of test)

# Review Manual Assignment

Ch. 11 and 12:

p. 73-75, Level 1 Q #22-34, Level 2 Q#2,7,8

Ch. 13-15:

p. 114-118 all Level 1 (22 total) and Level 2 (7 total)

Ch. 18.4: p. 148 #35

Ch. 47.3: p. 257 #35

- Dominance/recessiveness relationships have three important points.
- 1. They range from complete dominance, though various degrees of incomplete dominance, to codominance.
- 2. They reflect the mechanisms by which specific alleles are expressed in the phenotype and do not involve the ability of one allele to subdue another at the level of DNA.
- 3. They do not determine or correlate with the relative abundance of alleles in a population, Education, Inc., publishing as Benjamin Cummings

### Degrees of Dominance

- Complete dominance occurs when phenotypes of the heterozygote and dominant homozygote are identical
- In incomplete dominance, the phenotype of F<sub>1</sub>
  hybrids is somewhere between the phenotypes of
  the two parental varieties
- In codominance, two dominant alleles affect the phenotype in separate, distinguishable ways

## Multiple Alleles

- Most genes exist in populations in more than two allelic forms
- •For example, the four phenotypes of the ABO blood group in humans are determined by three alleles for the enzyme (I) that attaches A or B carbohydrates to red blood cells:  $I^A$ ,  $I^B$ , and i.
- •The enzyme encoded by the *I*<sup>A</sup> allele adds the A carbohydrate, whereas the enzyme encoded by the *I*<sup>B</sup> allele adds the B carbohydrate; the enzyme encoded by the *i* allele adds neither

## X-linked (Sex-linked) Diseases

•Remind yourself how to draw the Punnett square for these – what do you need first?

Warm-up: next slide Be sure kids can log into network

12/5 Put up key to Genetics Practice Problems pg. 2 - check answers Slides on last types of inheritance and pedigrees

Work on your own - 2 assignments: (I check Ch. 14 notes)

- 1) Continue Case study with the next handout
  - do all of Part III will need to read about Stem cells in Ch. 20. p. 415-417
- 2) Research tumor suppressor genes answer on binder paper
- Read pgs. 374-5 & answer "what do tumor suppressors do in the cell cycle?"
- Specific examples of mutated tumor suppressor genes that may lead to cancer include human papillomavirus (HPV), retinoblastoma protein(Rb), BRCA 1 and BRCA2, and p53. Split these up among your group members and create a written summary of your research findings.

#### Homework -

Genetics Practice Problems packet fully done for Monday

- Ch. 14 online assignment due Monday (long! 2hrs)
- Ch. 15 Notes, concept checks due next Thurs and online assignment due next Friday
- •Next Unit Test (Ch. 11-15, 18.4, 47.3) approx. 12/16, be working on review manual chapters 11-15 (will be collected day of test)

# Dihybrid Crosses – Using Probability – Rules of Multiplication and Addition

In a dihybrid cross of 2 yellow pea plants with round seeds, 64 offspring are produced. Approximately how many of these offspring will be: Yellow with round seeds? Yellow with wrinkled seeds? Green with round seeds? Green with wrinkled seeds?

- Dominance/recessiveness relationships have three important points.
- 1. They range from complete dominance, though various degrees of incomplete dominance, to codominance.
- 2. They reflect the mechanisms by which specific alleles are expressed in the phenotype and do not involve the ability of one allele to subdue another at the level of DNA.
- 3. They do not determine or correlate with the relative abundance of alleles in a population, Education, Inc., publishing as Benjamin Cummings

## **Pleiotropy**

- Most genes have multiple phenotypic effects, a property called pleiotropy
- •For example, pleiotropic alleles are responsible for the multiple symptoms of certain hereditary diseases, such as cystic fibrosis and sickle-cell disease

## **Epistasis**

#### More than one gene

- In epistasis, a gene at one locus alters the phenotypic expression of a gene at a second locus
- For example, in Labrador retrievers and many other mammals, coat color depends on two genes
- One gene determines the pigment color (with alleles B for black and b for brown)
- The other gene (with alleles C for color and c for no color) determines whether the pigment will be deposited in the hair

#### More than one gene

## Polygenic Inheritance

- Quantitative characters are those that vary in the population along a continuum
- •Quantitative variation usually indicates **polygenic inheritance**, an additive effect of two or more genes on a single phenotype
- Skin color in humans is an example of polygenic inheritance

#### NATURE VS. NURTURE

- Phenotype depends on environment and genes.
  - A single tree has leaves that vary in size, shape, and greenness, depending on exposure to wind and sun.
  - For humans, nutrition influences height, exercise alters build, sun-tanning darkens the skin, and experience improves performance on intelligence tests.
  - Even identical twins, genetic equals, accumulate phenotypic differences as a result of their unique experiences.
- •The relative importance of genes and the environment in influencing human characteristics is a very old and hotly contested debate.

# Nature and Nurture: The Environmental Impact on Phenotype

- Another departure from Mendelian genetics arises when the phenotype for a character depends on environment as well as genotype
- The norm of reaction is the phenotypic range of a genotype influenced by the environment
- For example, hydrangea flowers of the same genotype range from blue-violet to pink, depending on soil acidity

## Pedigree Analysis

- A pedigree is a family tree that describes the interrelationships of parents and children across generations
- Inheritance patterns of particular traits can be traced and described using pedigrees