What in the world?
Who’s got a guess?
Cyclopic lamb

At one time, 5-7% of newborn sheep in Utah and Idaho were cyclopic, a condition officially known as holoprosencephaly.
Cyclopic Kitten

It isn’t just a sheep thing.
You guessed it.

Fetal alcohol syndrome can include holoprosencephaly as a symptom. How???
It’s all about HOX genes

• The Hox gene Sonic Hedgehog is turned off by a chemical, cyclopamine, that is in flowers some momma sheep were eating.
• Alcohol can also disable this gene, causing the same effect.
• However!!! Cyclopamine is also now used to disable this gene in cancer cells that use it to do their bad cancer thing.
AP objective

• Student Objectives:
  • How do the events of meiosis explain the observations of Gregor Mendel?

• ON the next slide,
  • (a) is level 1
  • (b) 1 is level 2
  • (b) 2 is level 3
Essential knowledge 3.A.3: The chromosomal basis of inheritance provides an understanding of the pattern of passage (transmission) of genes from parent to offspring.

a. Rules of probability can be applied to analyze passage of single gene traits from parent to offspring.

b. Segregation and independent assortment of chromosomes result in genetic variation.

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

1. Segregation and independent assortment can be applied to genes that are on different chromosomes.

2. Genes that are adjacent and close to each other on the same chromosome tend to move as a unit; the probability that they will segregate as a unit is a function of the distance between them.
• The first person to have a correct idea of genetics worked in an abbey garden, where a monk named Gregor Mendel documented the particulate mechanism of inheritance.

• Just what does “particulate” mean, anyway?
1. A little Mendel history

- Mendel grew up on a small farm in what was then Austria and is today the Czech Republic.
- In 1843, he entered an Augustinian monastery.
- He studied at the University of Vienna from 1851 to 1853 where he was influenced by Doppler (who’s he?), who encouraged experimentation and the application of mathematics to science.
- He then taught high school for a while, but because of his vow of poverty, he couldn’t keep the huge bucks that teachers make.
• Around 1857, Mendel began breeding garden peas to study inheritance.

• Pea plants have several advantages for genetics.
  – Pea plants are available in many varieties with distinct heritable features with different variants (traits).
  – Mendel chose 7 traits to work with that were either/or types of traits.
Another advantage of peas is that Mendel could control which plants mated with which.

Each pea plant has male (stamens) and female (carpal) sexual organs.

In nature, pea plants typically self-fertilize, fertilizing ova with their own sperm.

However, Mendel could also move pollen from one plant to another to cross-pollinate plants, just like you did to fast plants with those poor bees.
In his classic experiments, Mendel would first develop **pure or true-breeding** pea varieties, which he called the **P generation**.

Then he would cross two different true-breeding parents, calling their **hybrid** offspring the **F₁ generation**.

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2. Results of P generation crosses:

- If a blending model were correct, the F₁ hybrids from a cross between purple-flowered and white-flowered pea plants would have pale purple flowers.

- Instead, the F₁ hybrids all have purple flowers, just as purple as the purple-flowered parents.

Fig. 14.2
Dominant - Recessive

• He got these results for each of the seven traits, and concluded what he called his **Law of Dominance** - the factor that caused one trait could completely mask the effect of the factor that caused the alternative trait.
• Then Mendel allowed the F<sub>1</sub> plants to self-fertilize to see what happened to the recessive trait factor.
• The recessive white trait, absent in the F<sub>1</sub>, reappeared in the F<sub>2</sub>, *always in a certain ratio*.
• Based on a large sample size, Mendel recorded 705 purple-flowered F<sub>2</sub> plants and 224 white-flowered F<sub>2</sub> plants from the original cross.
• This cross produced a three purple to one white ratio of traits in the F<sub>2</sub> offspring,

• The reappearance of white-flowered plants in the F<sub>2</sub> generation indicated that the inherited factor for the white trait was not diluted or “blended” by coexisting with the purple-flower factor in F<sub>1</sub> hybrids.
<table>
<thead>
<tr>
<th>Character</th>
<th>Dominant Trait</th>
<th>Recombinant Trait</th>
<th>$F_2$ Generation</th>
<th>Ratio</th>
</tr>
</thead>
<tbody>
<tr>
<td>Flower color</td>
<td>Purple</td>
<td>White</td>
<td>705:224</td>
<td>3.15:1</td>
</tr>
<tr>
<td>Flower position</td>
<td>Axial</td>
<td>Terminal</td>
<td>651:207</td>
<td>3.14:1</td>
</tr>
<tr>
<td>Seed color</td>
<td>Yellow</td>
<td>Green</td>
<td>6022:2001</td>
<td>3.01:1</td>
</tr>
<tr>
<td>Seed shape</td>
<td>Round</td>
<td>Wrinkled</td>
<td>5474:1850</td>
<td>2.96:1</td>
</tr>
<tr>
<td>Pod shape</td>
<td>Inflated</td>
<td>Constricted</td>
<td>882:299</td>
<td>2.95:1</td>
</tr>
<tr>
<td>Pod color</td>
<td>Green</td>
<td>Yellow</td>
<td>428:152</td>
<td>2.82:1</td>
</tr>
<tr>
<td>Stem length</td>
<td>Tall</td>
<td>Dwarf</td>
<td>787:277</td>
<td>2.84:1</td>
</tr>
</tbody>
</table>
Mendel developed a hypothesis to explain these 3 dominant:1 recessive results for phenotypes of the F$_2$ generation that consisted of four related ideas.

1. Alternative versions of genes (different alleles) account for variations in inherited traits. (He didn’t use “gene”).

2. For each trait, an organism has two factors, one inherited from each parent.

3. These factors may be identical, as in the true-breeding plants of the P generation.

4. Or the two factors may differ
   - In the flower-color example, the F$_1$ plants inherited a purple-flower factor from one parent and a white-flower factor from the other.
3. The two alleles for each trait **segregate** (separate) during gamete production.

- Since genes are a segment of a chromosome, this segregation of alleles corresponds to the distribution of homologous chromosomes to different gametes in meiosis I, **but Mendel knew nothing of this.**
  - If an organism has identical genes for a particular trait, then all the gametes they make will have that gene.
  - If different alleles are present, then 50% of the gametes will receive one allele and 50% will receive the other.

- The separation of alleles into separate gametes is summarized as Mendel’s **law of segregation**.
• Mendel’s law of segregation explains the 3:1 ratio that he observed in the F₂ generation.
• The F₁ hybrids will produce two classes of gametes, half with the purple-flower allele and half with the white-flower allele.
• During self-pollination, the gametes of these two classes unite randomly.
• This can produce four equally likely combinations of sperm and ovum.
• You will get the 3:1 results only if both parents can make equal amounts of different gametes.
• If heredity worked this way, Mendel reasoned, you would get the 3 dominant to every 1 recessive result in the $F_2$ generation that he did.

• A Punnett square predicts the results of a genetic cross between individuals with known genes.

Fig. 14.4
• Genetics has some unique, useful vocabulary.

• An organism with two identical alleles for a trait is **homozygous** for that trait. TT, tt, BB, rr, etc.

• Organisms with two different alleles for a trait is **heterozygous** for that trait. Tt, Bb, Rr, etc.

• A description of an organism’s traits is its **phenotype**. *Black* hair, *green* peas, etc.

• A description of its genetic makeup is its **genotype**.
  
  – Two organisms can have the same phenotype but have different genotypes if one is homozygous dominant and the other is heterozygous. TT and Tt cause the same trait.
• You can’t tell the genotype of an organism with a dominant phenotype.
  – The organism must have one dominant allele, but it could be homozygous dominant or heterozygous.

• A **test cross**, breeding a homozygous recessive with dominant phenotype, but unknown genotype, can determine the identity of the unknown allele.

Fig. 14.6
How about these symbols? Mistake here?
Essential knowledge 3.A.3: The chromosomal basis of inheritance provides an understanding of the pattern of passage (transmission) of genes from parent to offspring.

a. Rules of probability can be applied to analyze passage of single gene traits from parent to offspring.
b. Segregation and independent assortment of chromosomes result in genetic variation.

Evidence of student learning is a demonstrated understanding of each of the following:
1. Segregation and independent assortment can be applied to genes that are on different chromosomes.
Learning Objectives:

LO 3.12 The student is able to construct a representation that connects the process of meiosis to the passage of traits from parent to offspring. [See SP 1.1, 7.2]

LO 3.13 The student is able to pose questions about ethical, social or medical issues surrounding human genetic disorders. [See SP 3.1]

LO 3.14 The student is able to apply mathematical routines to determine Mendelian patterns of inheritance provided by data sets. [See SP 2.2]

Essential knowledge 3.A.4: The inheritance pattern of many traits cannot be explained by simple Mendelian genetics.

a. Many traits are the product of multiple genes and/or physiological processes.
3. By the law of independent assortment, each pair of alleles segregates into gametes independently

- Mendel’s experiments that followed the inheritance of flower color or other traits focused on only a single trait, so we call them **monohybrid** crosses.
- He conducted other experiments in which he followed the inheritance of two different traits, a **dihybrid** cross.
- Organisms have thousands of traits, of course. A cross is called mono- or dihybrid depending simply on how many traits you want to keep track of.
In a dihybrid cross experiment, Mendel tried to see if one trait affected another as they were passed on.

Observation suggests they may (red hair & freckles, eg.). Here’s an example of what he did:

- The allele for yellow seeds (Y) is dominant to the allele for green seeds (y).
- The allele for round seeds (R) is dominant to the allele for wrinkled seeds (r).

Mendel crossed true-breeding plants that had yellow, round seeds (YYRR) with true-breeding plants that had green, wrinkled seeds (yyrr).

This is the same basic thing he did to begin his monohybrid cross series, cross pure P generations.
• One possibility is that the two characters are transmitted from parents to offspring as a package.
  – The Y and R alleles and y and r alleles stay together.
• If this were the case, the F₁ offspring would produce yellow, round seeds.
• The F₂ offspring would produce two phenotypes in a 3:1 ratio, just like a monohybrid cross.
• This was not consistent with Mendel’s results.
• Another idea is that the two pairs of alleles have no effect on each other when they are passed down.
  – The presence in a gamete of one specific allele for one trait has no impact on the presence of a specific allele for the second trait.

• In our example, the F\(_1\) offspring would still produce yellow, round seeds.

• However, when the F\(_1\)’s produced gametes, genes would be packaged into gametes in all 4 possible combinations.
  – Four classes of gametes (YR, Yr, yR, and yr) would be produced in equal amounts.
• When sperm with four classes of alleles and ova with four classes of alleles combined, there would be 16 **equally probable** ways in which the alleles can combine in the $F_2$ generation.

• These combinations produce four distinct **phenotypes** in a **9:3:3:1** ratio.

• This **was** what Mendel got.
• Mendel repeated the dihybrid cross experiment for other pairs of characters and always observed a **9:3:3:1 phenotypic ratio in the F₂ generation**.

• Just as a 3 dominant: 1 recessive phenotype ratio in the F2’s of his monohybrid cross was the key clue to figuring out segregation, this was the key ratio.

• Each trait appeared to be inherited independently.

• The independent assortment of each pair of alleles during gamete formation is now called Mendel’s **law of independent assortment** (it would have made too much sense to call it independent segregation).

• Let’s try 5,6 and 7 on the Critical Thinking/More Mendel Problems worksheet.
Hey Coach!

• We are all dying to know how this independent assortment thing relates to what happens to chromosomes in meiosis. We would hardly be able to contain our excitement if you came to the board and did some of those great diagrams for us. We also promise to be able to explain it without using the words “independent” or “assortment” in our explanation.
4. Mendelian inheritance reflects rules of probability

• Mendel’s laws of segregation and independent assortment reflect the same laws of probability that apply to tossing coins or rolling dice.

• The probability scale ranges from zero (an event with no chance of occurring) to one (an event that is certain to occur).
  – The probability of tossing heads with a normal coin is $\frac{1}{2}$.
  – The probability of rolling a 3 with a six-sided die is $\frac{1}{6}$, and the probability of rolling any other number is $1 - \frac{1}{6} = \frac{5}{6}$. 

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• We can use the **product rule** to determine the chance that two or more independent events will occur together in some specific combination.
  – Compute the probability of each independent event.
  – Then, multiply the individual probabilities to obtain the overall probability of these events occurring together.
  – The probability that two coins tossed at the same time will land heads up is $1/2 \times 1/2 = 1/4$.
  – Similarly, the probability that two heterozygous pea plants (Pp x Pp) will produce a white-flowered offspring (pp) depends on a sperm with a white allele fertilizing an egg with a white allele.
  – This probability is $1/2 \times 1/2 = 1/4$. 
The product rule also applies to dihybrid crosses.

- For a heterozygous parent (YyRr) the probability of producing a YR gamete is $1/2 \times 1/2 = 1/4$.
- We can use this to predict the probability of a particular $F_2$ genotype without constructing a 16-part Punnett square.
- The probability that an $F_2$ plant will have a YYRR genotype from a heterozygous parent is $1/16$ (1/4 chance for a YR ovum and 1/4 chance for a YR sperm).
- BUT BE VERY CAREFUL, because sometimes you have to use the **sum rule**. For example, what’s the probability of throwing two dice and getting a five and a two? Do 17 and 18 on the Genetics Worksheet.
Now for some practice math

• First with probability
• Then some chi square stuff
Tips

- Grid LEFT to right
- Use the formula sheet
- Don’t round until the end
- Look at HOW the answer should be given “round to nearest...”

.123
The 1 is in the **tenths** place
The 2 is in the **hundreds** place
The 3 is in the **thousandths** place
Q6: Laws of Probability

• Calculate the probability of tossing three coins simultaneously and obtaining three heads. Express in fraction form.
Q6

• Probability of a heads is $\frac{1}{2}$
• Probability of heads AND a heads AND a heads

$\frac{1}{2} \times \frac{1}{2} \times \frac{1}{2} = \frac{1}{8}$
Section B: Extending Mendelian Genetics

1. The relationship between genotype and phenotype is rarely simple
• The heterozygous F₁ offspring of Mendel’s crosses always looked like one of the parental varieties because one allele was dominant to the other.

• However, some alleles show **incomplete dominance** where heterozygotes show a distinct intermediate phenotype, not seen in homozygotes.
  
  – This is not blended inheritance because the traits are separable (particulate) as seen in further crosses.
  
  – Offspring of a cross between heterozygotes will show three phenotypes: both parentals and the heterozygote.
  
  – The phenotypic and genotypic ratios are identical, 1:2:1.
A clear example of incomplete dominance is seen in flower color of snapdragons.

- A cross between a white-flowered plant and a red-flowered plant will produce all pink $F_1$ offspring.
- Self-pollination of the $F_1$ offspring produces 25% white, 25% red, and 50% pink offspring.
• Complete and incomplete dominance are part of a spectrum of relationships among alleles.

• At the other extreme from complete dominance is codominance in which two alleles affect the phenotype in separate, distinguishable ways.
  
  – For example, the ABO blood groups of humans are due to the presence of two specific molecules on the surface of red blood cells.

  – People of group A have one type of molecule on their red blood cells, people of group B have the other type, and people of group AB have both molecules present, not some kind of in-between molecule.

  – O type people have neither molecule present.
• Most genes have more than two alleles in a population.

• The ABO blood groups in humans are determined by three alleles, \( I^A, I^B, \) and \( i \).
  – Both the \( I^A \) and \( I^B \) alleles are dominant to the \( i \) allele
  – The \( I^A \) and \( I^B \) alleles are codominant to each other.

• Because each individual carries two alleles, there are six possible genotypes and four possible blood types (phenotypes).

• Many traits are like blood type in that they are controlled by multiple alleles.
— But while there may be many alleles for a trait, each person will only have a pair (that concept doesn’t change...yet).
— Individuals that are $I^A I^A$ or $I^A i$ are type A and place type A molecules on the surface of their red blood cells.
— Individuals that are $I^B I^B$ or $I^B i$ are type B and place type B molecules on the surface of their red blood cells.
— Individuals that are $I^A I^B$ are type AB and place both type A and type B molecules on the surface of their red blood cells. We say that both genes are “expressed”.
— Individuals that are $ii$ are type O and place neither molecule on the surface of their red blood cells.
— Let’s try 13, 14 and 16 on the Genetics Worksheet and 8,9,12 and 14 on the Cr. Th./More Mendel Worksheet
• Some traits do not fit the either-or basis that Mendel studied.

• These are usually due to **polygenic inheritance**, the additive effects of two or more gene pairs on a single phenotypic trait.
  
  – For example, skin color in humans is controlled by at least three different gene pairs.

  – Imagine that each gene has two alleles, one light and one dark, that demonstrate incomplete dominance.

  – An AABBCCC individual is the darkest and aabbcc is the lightest. Try 17 on the Cr. Th./ More Mendel wksht.
A cross between two AaBbCc individuals (intermediate skin shade) would produce offspring covering a wide range of shades.

- Individuals with intermediate skin shades would be the most likely offspring, but very light and very dark individuals are possible as well.
- The range of phenotypes forms a normal distribution.

Fig. 14.12
• 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, even more so today.
• In epistasis, a gene at one locus alters the phenotypic expression of a gene at a second locus.

– For example, in mice and many other mammals, coat color depends on two genes.

– One, the epistatic gene, determines whether pigment will be deposited in hair or not.
  • Presence (C) is dominant to absence (c).

– The second determines whether the pigment to be deposited is black (B) or brown (b).
  • The black allele is dominant to the brown allele.

– An individual that is cc has a white (albino) coat regardless of the genotype of the second gene.
• A cross between two black mice that are heterozygous (BbCc) will follow the law of independent assortment.

• However, unlike the 9:3:3:1 offspring ratio of a normal Mendelian experiment, the ratio is nine black, three brown, and four white.

• Try 13, 15 and 16 on the Cr.Th./MoreMendel
1. Pedigree analysis reveals Mendelian patterns in human inheritance

• Rather than manipulate mating patterns of people (a bit impractical and unethical), geneticists analyze the results of matings that have already occurred.

• In a **pedigree analysis**, information about the presence/absence of a particular phenotypic trait is collected from as many individuals in a family as possible and across generations.

• The distribution of these characters is then mapped on the family tree.
For example, the occurrence of widows peak (W) is dominant to a straight hairline (w).

The relationship among alleles can be integrated with the phenotypic appearance of these traits to predict the genotypes of members of this family.
• For example, if an individual in the third generation lacks a widow’s peak, but both her parents have widow’s peaks, then her parents must be heterozygous for that gene.

• If some siblings in the second generation lack a widow’s peak and one of the grandparents (first generation) also lacks one, then we know the other grandparent must be heterozygous and we can determine the genotype of almost all other individuals. Notice the colored symbol is the dominant trait.
– We can use the same family tree to trace the distribution of attached earlobes (f), a recessive characteristic.

– **Notice the colored-in trait is the recessive one here.**

– Individuals with a dominant allele (F) have free earlobes.

– Some individuals may be ambiguous, especially if they have the dominant phenotype and could be heterozygous or homozygous dominant. So let’s practice.
c. Certain human genetic disorders can be attributed to the inheritance of single gene traits or specific chromosomal changes, such as nondisjunction.

*To demonstrate your understanding, make sure you can explain examples like:*

- Sickle cell anemia
- Tay-Sachs disease
- Huntington’s disease
- X-linked color blindness
- Trisomy 21/Down syndrome
- Klinefelter’s syndrome
A lethal, recessive disease is **cystic fibrosis** which strikes one of every 2,500 whites of European descent.

- One in 25 whites is a carrier.
- The normal allele codes for a membrane protein that transports $\text{Cl}^-$ between cells and the environment.
- If these channels are defective or absent, there are abnormally high extracellular levels of chloride that causes the mucus coats of certain cells to become thicker and stickier than normal.
- This mucus build-up in the pancreas, lungs, digestive tract, and elsewhere favors bacterial infections.
- Without treatment, affected children die before five, but with treatment can live past their late 20’s.
• Tay-Sachs disease is another lethal recessive disorder.
  – It is caused by a dysfunctional enzyme that fails to break down specific brain lipids.
  – The symptoms begin with seizures, blindness, and degeneration of motor and mental performance a few months after birth.
  – Inevitably, the child dies after a few years.
  – Among Ashkenazic Jews (those from central Europe) this disease occurs in one of 3,600 births, about 100 times greater than the incidence among non-Jews or Mediterranean (Sephardic) Jews. DNA studies have shown that they all descended from one of 4 women.
The most common inherited disease among blacks is **sickle-cell disease**.

- It affects one of 400 African Americans.
- It is caused by the substitution of a single amino acid in hemoglobin.
- When oxygen levels in the blood of an affected individual are low, sickle-cell hemoglobin crystallizes into long rods.
- This deforms red blood cells into a sickle shape.
• This sickling creates a cascade of symptoms, demonstrating the pleiotropic effects of this allele.

• Doctors can use regular blood transfusions to prevent brain damage and new drugs to prevent or treat other problems.

Fig. 14.15
• At the organismal level, the non-sickle allele is incompletely dominant to the sickle-cell allele.
  – Carriers are said to have the sickle-cell trait.
  – These individuals are usually healthy, although some suffer some symptoms of sickle-cell disease under blood oxygen stress.
• At the molecule level, the two alleles are codominant as both normal and abnormal hemoglobins are synthesized.
• So let’s watch an HHMI presentation on the discovery of a connection between sickle cell and malaria. Then a worksheet!
• The high frequency of heterozygotes with the sickle-cell trait is unusual for an allele with severe detrimental effects in homozygotes.
  – Interestingly, individuals with one sickle-cell allele have increased resistance to malaria, a parasite that spends part of its life cycle in red blood cells.
  – In tropical Africa, where malaria is common, the sickle-cell allele is both a boon and a bane.
    • Homozygous normal individuals die of malaria, homozygous recessive individuals die of sickle-cell disease, and carriers are relatively free of both.

• Its relatively high frequency in African Americans is a vestige of their African roots.
• Normally it is relatively unlikely that two carriers of the same rare harmful allele will meet and mate.
• However, consanguineous matings, those between close relatives, increase the risk.
  – These individuals who share a recent common ancestor are more likely to carry the same recessive alleles.
• Most societies and cultures have laws or taboos forbidding marriages between close relatives.
• Queen Victoria and family (hemophilia), and the Hapsburgs (jaw deformity and insanity) are famous examples of inbreeding gone wild...
These dogs, obviously are inbred
Please...
NEUTER YOUR PETS
AND
WEIRD FRIENDS & RELATIVES
• Although most harmful alleles are recessive, many human disorders are due to dominant alleles.
• For example, *achondroplasia*, a form of dwarfism, has an incidence of one case in 10,000 people.
  – Heterozygous individuals have the dwarf phenotype.
  – Those who are not achondroplastic dwarfs, 99.99% of the population are homozygous recessive for this trait.
• Lethal dominant alleles are much less common than lethal recessives because if a lethal dominant kills an offspring before it can mature and reproduce, the allele will not be passed on to future generations.
• A lethal dominant allele can escape elimination if it causes death at a relatively advanced age, after the individual has already passed on the lethal allele to his or her children.

• One example is **Huntington’s disease**, a degenerative disease of the nervous system.
  – The dominant lethal allele has no obvious phenotypic effect until an individual is about 35 to 45 years old.
  – The deterioration of the nervous system is irreversible and inevitably fatal.
• Any child born to a parent who has the allele for Huntington’s disease has a 50% chance of inheriting the disease and the disorder.

• Recently, molecular geneticists have used pedigree analysis of affected families to track down the Huntington’s allele to a locus near the tip of chromosomes 4.

Fig. 14.15
While some diseases are inherited in a simple Mendelian fashion due to alleles at a single locus, many other disorders have a multifactorial basis.

- These have a genetic component plus a significant environmental influence.
- Multifactorial disorders include heart disease, diabetes, cancer, alcoholism, and certain mental illnesses, such as schizophrenia and manic-depressive disorder.
- The genetic component is typically polygenic.

At present, little is understood about the genetic contribution to most multifactorial diseases
- The best public health strategy is education about the environmental factors and healthy behavior.
3. Technology is providing new tools for genetic testing and counseling

- A preventative approach to simple Mendelian disorders is sometimes possible.
- The risk that a particular genetic disorder will occur can sometimes be assessed before a child is conceived or early in pregnancy.
- Many hospitals have genetic counselors to provide information to prospective parents who are concerned about a family history of a specific disease.
• Tests are also available to determine in utero if a child has a particular disorder.
• One technique, amniocentesis, can be used beginning at the 14\textsuperscript{th} to 16\textsuperscript{th} week of pregnancy to assess the presence of a specific disease.
  – Fetal cells extracted from amniotic fluid are cultured and karyotyped to identify some disorders.
A second technique, chorionic villus sampling (CVS) can allow faster karyotyping and can be performed as early as the eighth to tenth week of pregnancy.

- This technique extracts a sample of fetal tissue from the chorionic villi of the placenta.

- This technique is not suitable for tests requiring amniotic fluid.

Fig. 14.17b
• Other techniques, *ultrasound* and *fetoscopy*, allow fetal health to be assessed visually in utero.

• Both fetoscopy and amniocentesis cause complications in about 1% of cases.
  – These include maternal bleeding or fetal death.
  – Therefore, these techniques are usually reserved for cases in which the risk of a genetic disorder or other type of birth defect is relatively great.

• If fetal tests reveal a serious disorder, the parents face the difficult choice of terminating the pregnancy or preparing to care for a child with a genetic disorder.
• Some genetic tests can be detected at birth by simple tests that are now routinely performed in hospitals.
• One test can detect the presence of a recessively inherited disorder, phenylketonuria (PKU).
  – This disorder occurs in one in 10,000 to 15,000 births.
  – Individuals with this disorder accumulate the amino acid phenylalanine and its derivative phenylpyruvate in the blood to toxic levels.
  – This leads to mental retardation.
  – If the disorder is detected, a special diet low in phenylalanine usually promotes normal development.