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his offspring.

- Because the **mother** has an affected sibling and neither of her parents is affected, she most likely had 2 heterozygous carrier parents. Therefore, the mother's 4 possible genotypes are: homozygous for the normal allele, heterozygous with her mother's mutant allele, heterozygous with her father's mutant allele, and homozygous for the mutant allele. However, the mother does **not have CF** and therefore is **not homozygous for the mutant allele**. This leaves 3 possible genotypes for the mother. Two of the 3 remaining genotypes result in her being a **carrier** for the mutant *CFTR* allele, while the last one results in her being homozygous normal. Therefore, the mother's probability of being a carrier equals **2/3**.
- If the mother is a carrier (2/3 chance), the probability that she will transmit the mutant allele to the child is 1 in 2. As a result, the probability that the child will inherit a mutant allele from the mother (and therefore have CF as the father will always contribute a mutant allele) is: $2/3 \times 1/2 = 1/3$.

Educational objective:

The probability that an autosomal recessive disease will be transmitted to a child can be calculated based on the maternal and paternal pedigrees. An unaffected individual (with unaffected parents) who has a sibling affected by an autosomal recessive condition has a 2/3 chance of being a carrier for that condition.

References

- Genetic counseling issues in cystic fibrosis.
- Cystic fibrosis and fertility.

Genetics
Subject

Genetics (General Principles)
System

Genetic inheritance
Topic

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Item 19 of 20 Question Id: 701

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A married couple comes to the office for routine prenatal counseling. The husband is 120 cm (3 ft 11 in) tall with disproportionately short upper and lower extremities, a large head, and a prominent forehead. He cannot provide a biological family history because he was adopted. His spouse is average height with no dysmorphic features, and her family history is insignificant. They inquire about the likelihood that their offspring will have short stature due to the same condition as the father. Which of the following is the best response to their concerns?

- A. The condition is not hereditary
- B. The probability depends on the child's biological sex
- C. The probability depends on the mother's carrier status
- D. The probability is about 25%
- E. The probability is about 50%

Omitted
Correct answer
E

Collecting Statistics

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Explanation

Autosomal dominant inheritance

Affected parent (Aa)

A a

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Autosomal dominant inheritance

Affected parent (Aa)

		A	a
		Aa Affected child	aa Normal child
a	A	Aa Affected child	aa Normal child
	a	Aa Affected child	aa Normal child

Offspring have 50% chance of being affected

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This man's short stature and morphologic features are suggestive of [achondroplasia](#), the most common form of bone dysplasia. It is caused by a **gain-of-function mutation** in the fibroblast growth factor receptor 3 (*FGFR3*) gene, which encodes a protein that inhibits chondrocyte proliferation and endochondral bone growth.

Achondroplasia occurs as a sporadic mutation in 85% of cases; advanced paternal age is a risk factor due to increased DNA replication errors during spermatogenesis. However, once a mutation occurs, it can be transmitted as an **autosomal dominant** trait (responsible for the remaining 15% of cases) with complete penetrance (Choice A).

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This man's short stature and morphologic features are suggestive of [achondroplasia](#), the most common form of bone dysplasia. It is caused by a **gain-of-function mutation** in the fibroblast growth factor receptor 3 (*FGFR3*) gene, which encodes a protein that inhibits chondrocyte proliferation and endochondral bone growth.

Achondroplasia occurs as a sporadic mutation in 85% of cases; advanced paternal age is a risk factor due to increased DNA replication errors during spermatogenesis. However, once a mutation occurs, it can be **transmitted as an autosomal dominant trait** (responsible for the remaining 15% of cases) with complete penetrance (**Choice A**).

Inheritance of only **1 mutant allele causes the disorder**; inheritance of 2 mutant alleles (ie, homozygous achondroplasia) is lethal. As a result, the **man must be heterozygous** for the achondroplasia mutation. A heterozygous parent has a 50% chance of transmitting the mutated allele; therefore, the unborn child has a **50% chance** of inheriting achondroplasia. Because achondroplasia is a rare condition, the probability of having a sporadic mutation does not significantly add to the probability of inheriting the disease.

(Choice B) In sex-linked disorders, the responsible gene is located on a sex chromosome (either X or Y). Most sex-linked disorders are [X-linked recessive](#); females with 1 copy of the defective gene will not have the disorder (they will be carriers), but all males who inherit the defective gene will be affected. In [X-linked dominant](#) disorders, both males and females are affected following inheritance of the defective gene.

(Choice C) Because inheritance of only 1 mutant allele causes achondroplasia and the disease is completely penetrant, carriers do not exist. This woman must be homozygous for the normal allele (since she is average height with no features that suggest achondroplasia).

(Choice D) About 25% of children are affected in [autosomal recessive](#) disorders if both parents carry 1 copy of the defective gene. Many recessive disorders are the result of enzymatic deficiencies (eg, phenylketonuria) that

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achondroplasia is lethal. As a result, the man must be ~~homozygous~~ heterozygous for the achondroplasia mutation.

A heterozygous parent has a 50% chance of transmitting the mutated allele; therefore, the unborn child has a **50% chance** of inheriting achondroplasia. Because achondroplasia is a rare condition, the probability of having a sporadic mutation does not significantly add to the probability of inheriting the disease.

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(Choice C) Because inheritance of only 1 mutant allele causes achondroplasia and the disease is completely penetrant, carriers do not exist. This woman must be homozygous for the normal allele (since she is average height with no features that suggest achondroplasia).

(Choice D) About 25% of children are affected in **autosomal recessive** disorders if both parents carry 1 copy of the defective gene. Many recessive disorders are the result of enzymatic deficiencies (eg, phenylketonuria) that require both copies of the gene to be knocked out, as 1 functional copy usually provides sufficient enzymatic activity to prevent occurrence of the disease.

Educational objective:

Achondroplasia is an autosomal dominant (AD) disorder that results in a gain-of-function mutation in the FGFR3 gene. Most individuals affected by AD disorders are heterozygous and have a 50% chance of transmitting the mutation to their offspring.

Genetics
Subject

Genetics (General Principles)
System

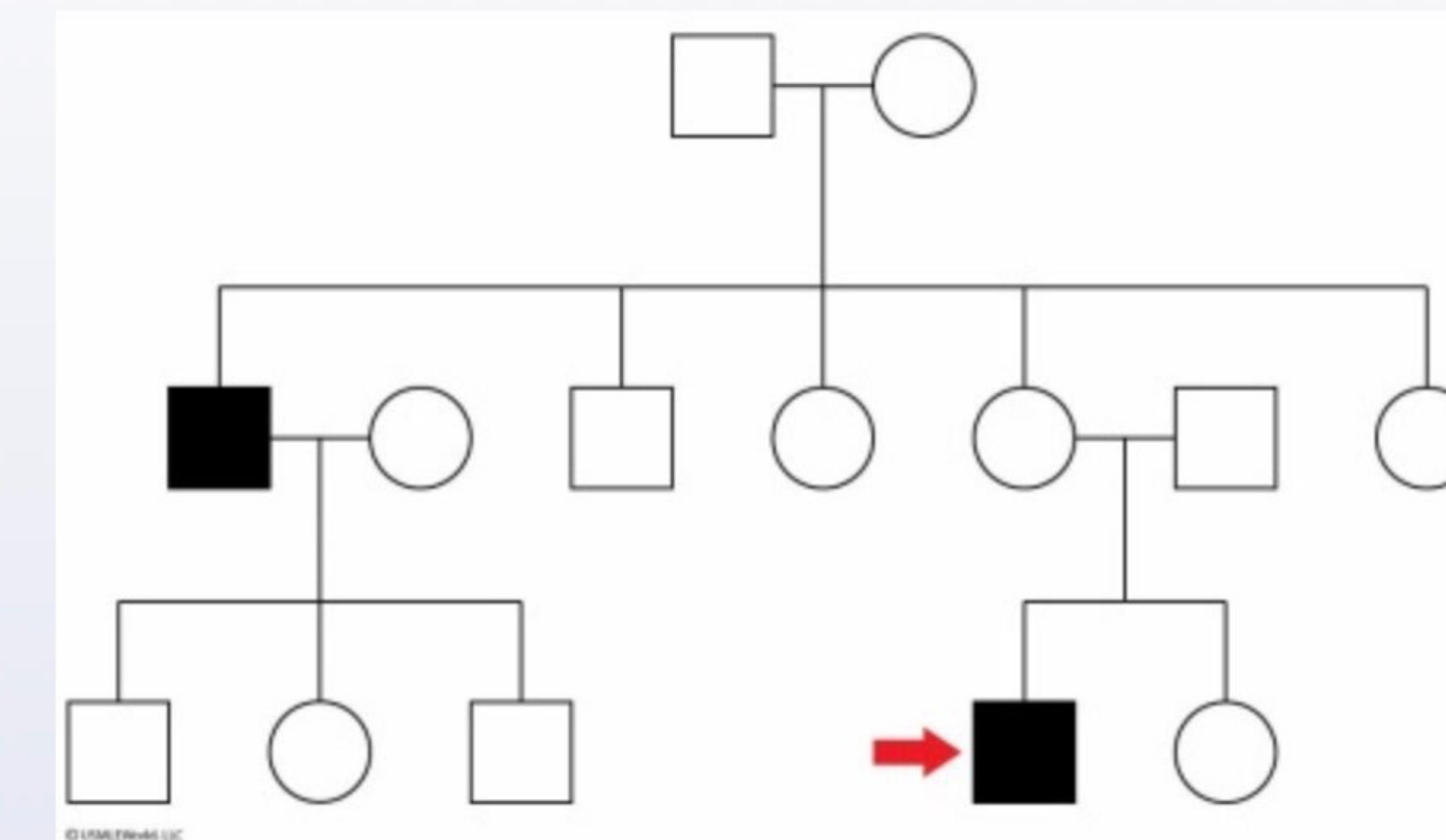
Achondroplasia
Topic

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A 25-year-old man experiences severe intolerance to certain medications. On 2 occasions, his reactions to various drugs have necessitated hospital admission. His family pedigree with respect to this condition is shown below, with the red arrow indicating his position within the family.



Assuming that the genetic condition demonstrates complete penetrance and is rare in the general population, which of the following inheritance patterns is most likely?

- A. Autosomal dominant (1%)
- B. Autosomal recessive (15%)
- C. X-linked dominant (4%)
- D. X-linked recessive (75%)
- E. Mitochondrial (3%)

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X-linked recessive inheritance

Affected father

		Mother		
		X	X	
Father	X ^d	XX ^d	XX ^d	All daughters are carriers
	Y	XY	XY	All sons are unaffected

Carrier mother

		Mother		
		X	X ^d	
Father	X	XX	XX ^d	Daughters have 50% chance of becoming carriers
	Y	XY	X ^d Y	Sons have 50% chance of being affected

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The pedigree shows that only males are affected by the drug intolerance. Specifically, **male offspring of unaffected parents** are affected. There is no evidence of male-to-male transmission. This pattern is most consistent with X-linked recessive inheritance from an asymptomatic carrier female in the first generation. In **X-linked recessive inheritance**:

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The pedigree shows that only males are affected by the drug intolerance. Specifically, **male offspring of unaffected parents** are affected. There is no evidence of male-to-male transmission. This pattern is most consistent with X-linked recessive inheritance from an asymptomatic carrier female in the first generation. In **X-linked recessive inheritance**:

1. Affected males will always produce unaffected sons and carrier daughters.
2. Carrier females have a 50% chance of producing an affected son or carrier daughter.

G6PD deficiency, which causes acute hemolytic anemia on exposure to oxidant drugs, follows an X-linked recessive pattern of inheritance.

(Choice A) With autosomal dominant inheritance, affected individuals have at least 1 parent that is also affected.

(Choice B) In autosomal recessive inheritance, both parents must at least be carriers (heterozygous) for the mutation in order to produce affected offspring. However, the father of the marked individual comes from *outside the family* and would be unlikely to carry the mutation (the question states the mutation is rare in the general population).

(Choice C) In **X-linked dominant inheritance**, affected individuals have at least 1 parent (of either sex) that is also affected. An affected male will always produce affected daughters, but none of his sons will be affected.

(Choice E) Conditions with mitochondrial inheritance are transmitted only by females. All of an affected female's offspring are likely to show signs of the disease.

Educational objective:

In X-linked recessive inheritance 1) affected males will always produce *unaffected* sons and *carrier* daughters, and 2) carrier females have a 50% chance of producing *affected* sons and *carrier* daughters. G6PD deficiency

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linked recessive inheritance:

1. Affected males will always produce unaffected sons and carrier daughters.
2. Carrier females have a 50% chance of producing an affected son or carrier daughter.

G6PD deficiency, which causes acute hemolytic anemia on exposure to oxidant drugs, follows an X-linked recessive pattern of inheritance.

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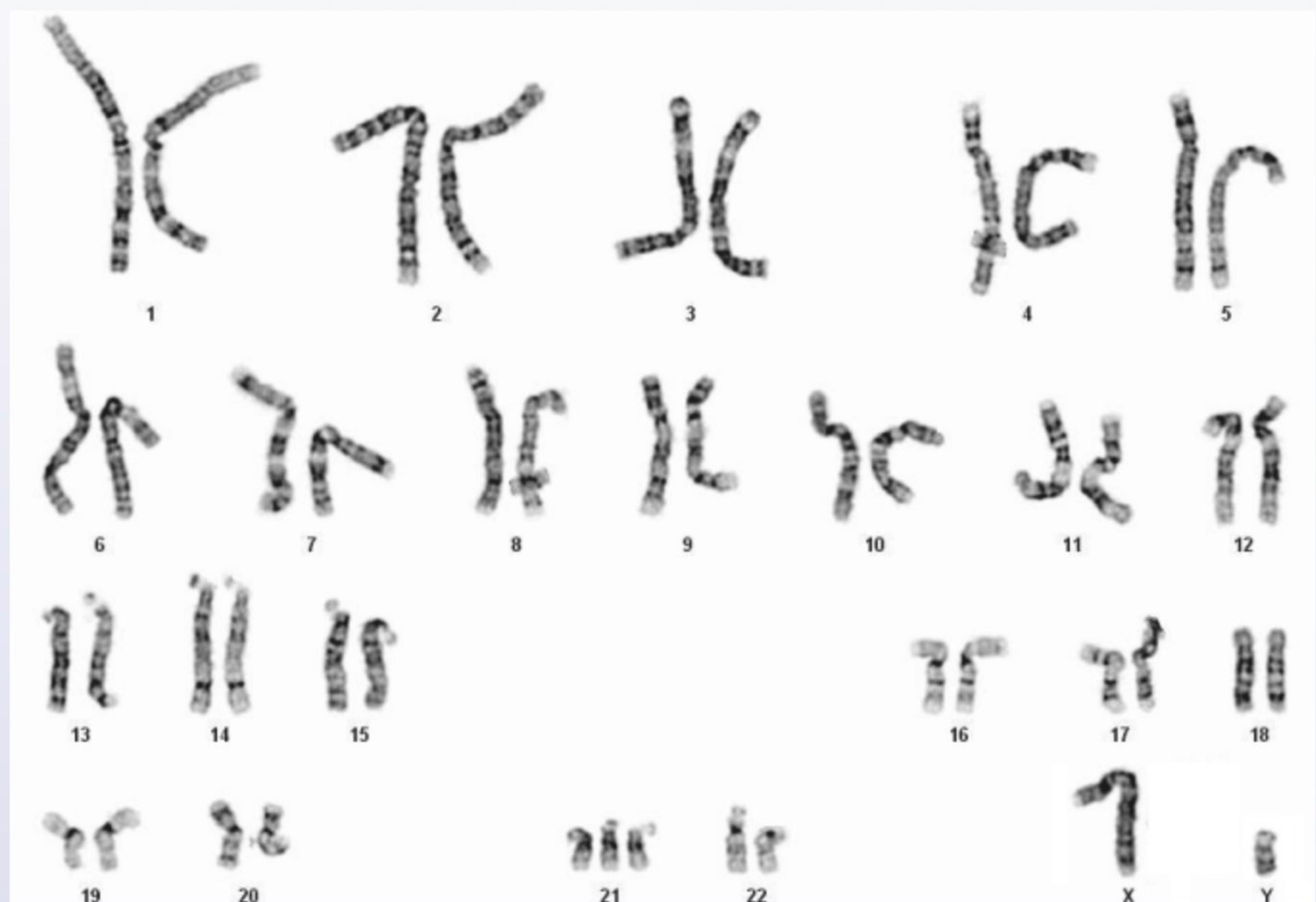
Educational objective:

In X-linked recessive inheritance 1) affected males will always produce *unaffected* sons and *carrier* daughters, and 2) carrier females have a 50% chance of producing *affected* sons and *carrier* daughters. G6PD deficiency follows this inheritance pattern and causes acute hemolytic anemia in response to oxidant drugs.

Miscellaneous

X

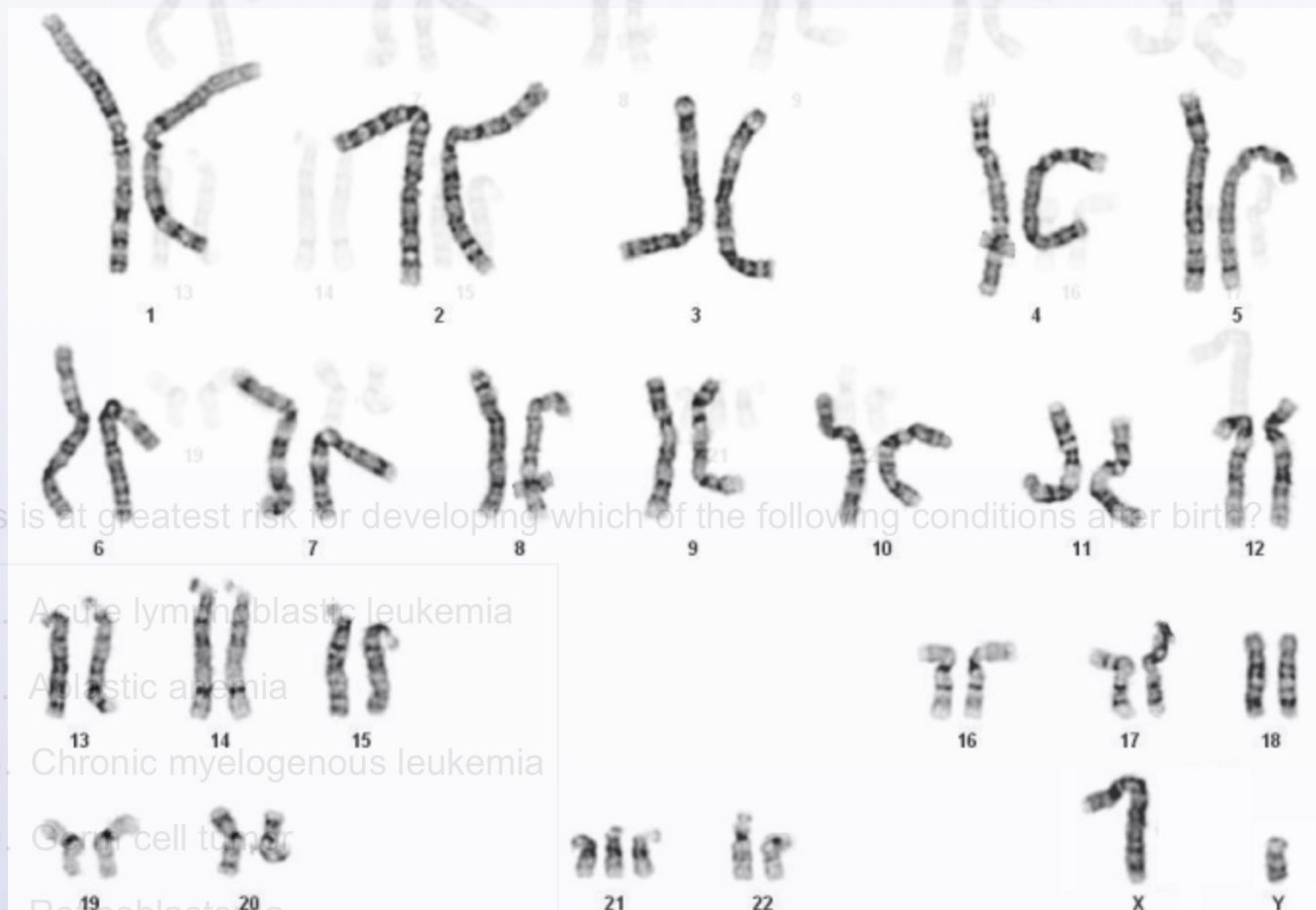
A 35-year-old woman, gravida 1 para 0, at 20 weeks gestation comes to the office for a routine prenatal visit and fetal anatomy ultrasound. The ultrasound reveals several abnormalities. An amniocentesis is performed and a fetal karyotype analysis is ordered; the results are shown in the image below:



This fetus is at greatest risk for developing which of the following conditions after birth?

- A. Acute lymphoblastic leukemia
- B. Aplastic anemia

A 35-year-old woman, gravida 1 para 0, at 20 weeks gestation comes to the office for a routine prenatal visit and fetal anatomy ultrasound. The ultrasound reveals several abnormalities. An amniocentesis is performed and a fetal karyotype analysis is ordered; the results are shown in the image below:



This fetus is at greatest risk for developing which of the following conditions after birth?

- A. Acute lymphoblastic leukemia
- B. Aplastic anemia
- C. Chronic myelogenous leukemia
- D. Granul cell tumor
- E. Retinoblastoma

This fetus is at greatest risk for developing which of the following conditions after birth?

- F. Rhabdomyosarcoma

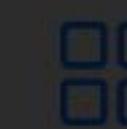
- A. Acute lymphoblastic leukemia
- B. Aplastic anemia

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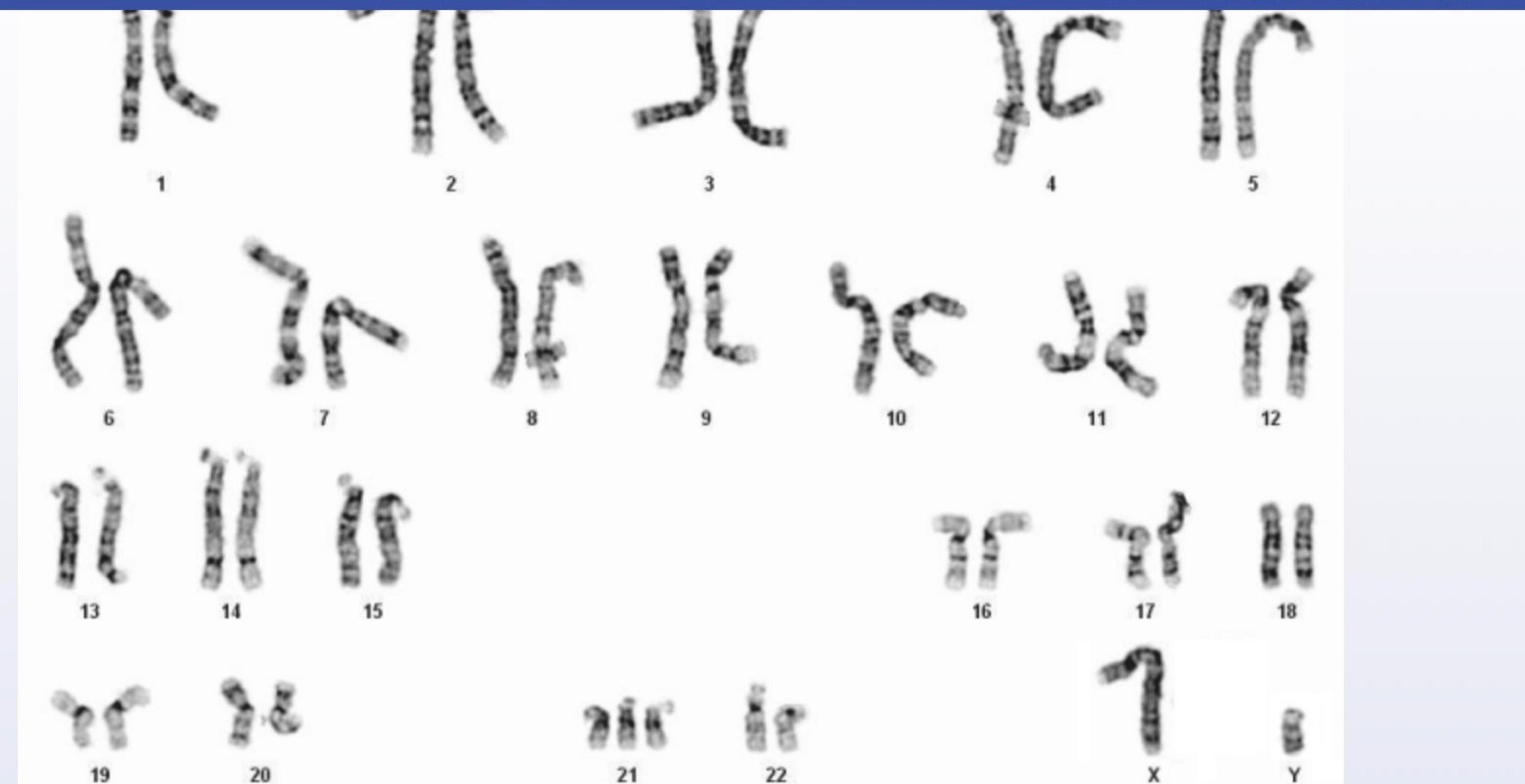
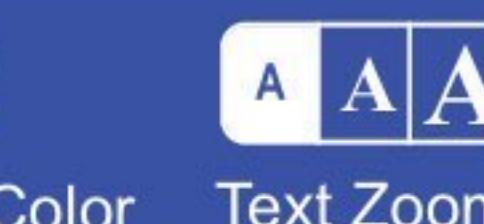
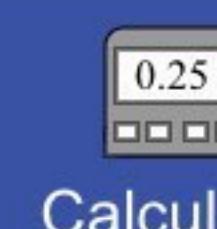
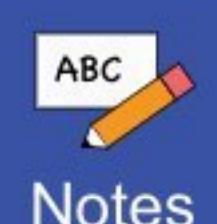
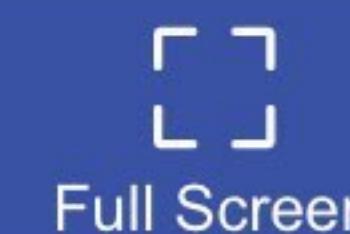


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This fetus is at greatest risk for developing which of the following conditions after birth?

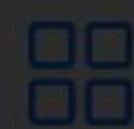
- A. Acute lymphoblastic leukemia
- B. Aplastic anemia
- C. Chronic myelogenous leukemia
- D. Germ cell tumor
- E. Retinoblastoma
- F. Rhabdomyosarcoma



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A man and woman come to the clinic for preconception counseling. The woman and her parents are healthy, but she has 2 siblings with a rare autosomal recessive disease. The woman's genotype is unknown. The man is also healthy and has no family history of the disease. It is known that, among healthy individuals in the general population, the carrier frequency is 1 in 200. What is the probability of this couple conceiving a child with the disease?

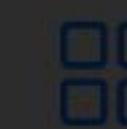
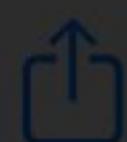
- A. 1/200
- B. 1/400
- C. 1/800
- D. 1/1,200
- E. 1/1,600

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Molecular biologists undertake a series of experiments designed to classify proteins involved in various intracellular signaling pathways. During one of the experiments, a protein mixture obtained from a cell culture is separated by gel electrophoresis and subsequently transferred to a filter membrane. Labeled double-stranded DNA probes are then used to detect a specific protein of interest in the sample. Which of the following proteins is most likely to be detected by this method?

- A. Ras
- B. c-Jun
- C. β 1-adrenoreceptor
- D. S-100
- E. Adenylate cyclase

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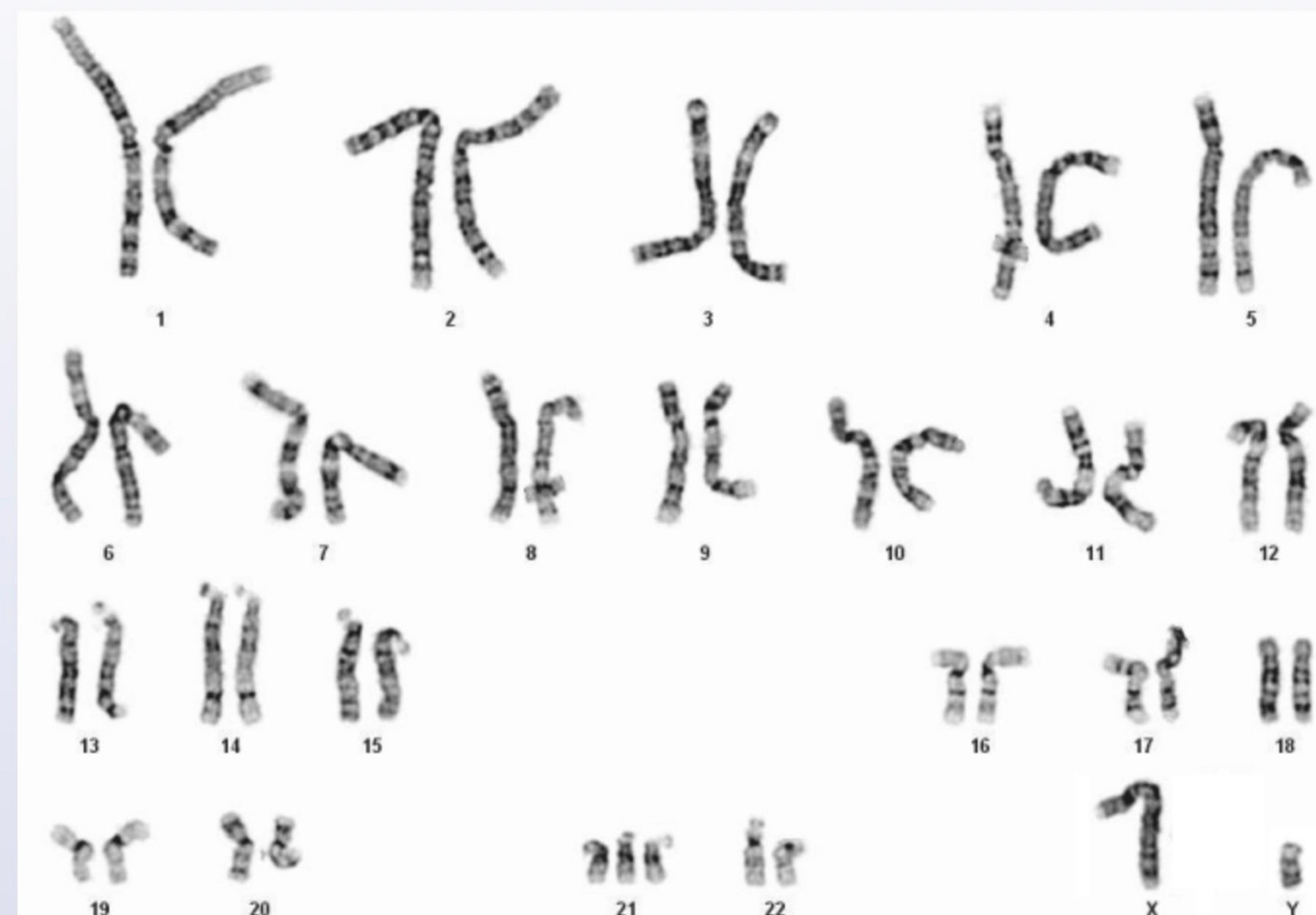
Calculator

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Text Zoom

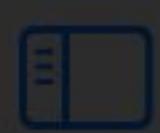
Settings

A 35-year-old woman, gravida 1 para 0, at 20 weeks gestation comes to the office for a routine prenatal visit and fetal anatomy ultrasound. The ultrasound reveals several abnormalities. An amniocentesis is performed and a fetal karyotype analysis is ordered; the results are shown in the image below:



This fetus is at greatest risk for developing which of the following conditions after birth?

- A. Acute lymphoblastic leukemia (85%)
- B. Aplastic anemia (1%)



AA



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20



21



X

Y

This fetus is at greatest risk for developing which of the following conditions after birth?

- A. Acute lymphoblastic leukemia (85%)
- B. Aplastic anemia (1%)
- C. Chronic myelogenous leukemia (7%)
- D. Germ cell tumor (1%)
- E. Retinoblastoma (2%)
- F. Rhabdomyosarcoma (1%)

Omitted

Correct answer
A85%
Answered correctly34 secs
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Version

Explanation

Down syndrome comorbidities	
Neurologic	<ul style="list-style-type: none">Intellectual disabilityEarly-onset Alzheimer disease
Cardiac	<ul style="list-style-type: none">Complete atrioventricular septal defectVentricular septal defect



AA



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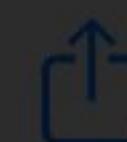
Down syndrome comorbidities	
Neurologic	<ul style="list-style-type: none">• Intellectual disability• Early-onset Alzheimer disease
Cardiac	<ul style="list-style-type: none">• Complete atrioventricular septal defect• Ventricular septal defect• Atrial septal defect
Gastrointestinal	<ul style="list-style-type: none">• Duodenal atresia• Hirschsprung disease
Endocrine	<ul style="list-style-type: none">• Hypothyroidism• Type 1 diabetes mellitus• Obesity• Short stature
Oncologic	<ul style="list-style-type: none">• Acute leukemia
Orthopedic	<ul style="list-style-type: none">• Atlantoaxial instability

This karyotype shows [trisomy 21](#) (47, XY, +21), which is diagnostic for [Down syndrome](#). This condition usually results from meiotic nondisjunction in the ovum, with advanced maternal age (age ≥ 35) as a risk factor. Prenatal diagnosis is often suspected based on associated ultrasound findings, including a thickened nuchal fold, **cardiac septal defects**, and/or gastrointestinal abnormalities (eg, **duodenal atresia**).

Down syndrome is the most common genetic cause of intellectual disability. In addition, Down syndrome significantly increases the risk of childhood **hematologic malignancies**, including [acute lymphoblastic](#)



AA



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Down syndrome is the most common genetic cause of intellectual disability. In addition, Down syndrome significantly increases the risk of childhood **hematologic malignancies**, including **acute lymphoblastic leukemia** (most common leukemia in patients with Down syndrome) and acute megakaryoblastic leukemia. The pathogenesis of the increased leukemia risk is uncertain, but may involve increased chromosome 21 gene expression promoting overall genomic instability.

(Choice B) Patients with Fanconi anemia, not Down syndrome, develop bone marrow failure (aplastic anemia) due to an inherited mutation that causes defective DNA repair. Karyotype analysis would be normal in this condition.

(Choice C) Chronic myelogenous leukemia is commonly associated with a reciprocal translocation between the long arms of chromosomes 9 and 22 (ie, Philadelphia chromosome). This translocation fuses the *BCR* gene on chromosome 22 to the *ABL* gene on chromosome 9, resulting in formation of the oncogenic *BCR-ABL* fusion gene. Unlike this case, karyotype analysis would show elongation of chromosome 9 and shortening of chromosome 22.

(Choice D) [Turner syndrome \(45, XO\)](#) and [Klinefelter syndrome \(47, XXY\)](#) increase the risk of ovarian germ cell tumors and extragonadal germ cell tumors, respectively. This patient's karyotype is inconsistent with these diagnoses, and germ cell tumors are not increased in patients with Down syndrome.

(Choices E and F) Incidence of retinoblastoma, which is associated with a retinoblastoma (*RB1*) gene mutation, and rhabdomyosarcoma is not increased in patients with Down syndrome.

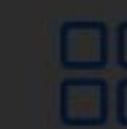
Educational objective:

Trisomy 21 (which is diagnostic for Down syndrome) is detectable by cytogenetic karyotype analysis. Patients are at increased risk of developing hematologic malignancies, including acute lymphoblastic leukemia and acute megakaryoblastic leukemia.



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AA



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Settings

A man and woman come to the clinic for preconception counseling. The woman and her parents are healthy, but she has 2 siblings with a rare autosomal recessive disease. The woman's genotype is unknown. The man is also healthy and has no family history of the disease. It is known that, among healthy individuals in the general population, the carrier frequency is 1 in 200. What is the probability of this couple conceiving a child with the disease?

- A. 1/200
- B. 1/400
- C. 1/800
- D. 1/1,200
- E. 1/1,600

Omitted

Correct answer

D

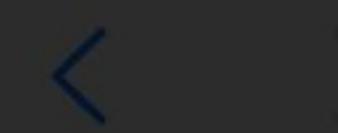
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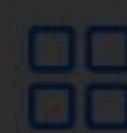
Explanation

For **autosomal recessive** disorders, an affected child must inherit a mutant allele from each parent; for 2 healthy parents to conceive an affected child (aa), they must each be a carrier for the disease (Aa). The probability of conceiving an affected child is equal to the **probability of both parents being carriers** multiplied by the **probability of each parent transmitting** the mutant allele:

$$P(\text{affected child}) = P(\text{carrier mother}) \times P(\text{carrier father}) \times P(\text{both parents transmit})$$



AA



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A A A



$P(\text{affected child}) = P(\text{carrier mother}) \times P(\text{carrier father}) \times P(\text{both parents transmitting a mutant allele})$

- $P(\text{carrier mother})$: The **woman has siblings affected** (aa) by the disease; because the **woman's parents are not affected** (ie, not aa), both of the woman's parents must be **carriers (Aa)**. This allows for the following genotypes in the woman:

- Homozygous mutant
- Heterozygous carrier with her mother's mutant allele
- Heterozygous carrier with the father's mutant allele
- Homozygous normal

However, because the woman is healthy, she **cannot be homozygous** for the mutant allele; she must have one of the **3 remaining genotypes**:

- Heterozygous carrier with her mother's mutant allele
- Heterozygous carrier with the father's mutant allele
- Homozygous normal

Therefore, the woman has a **2 in 3** probability of being a carrier.

- $P(\text{carrier father})$: The man's genotype is unknown and he has **no relevant family history**; therefore, the probability of him being a carrier is equal to the carrier frequency in the general population, or **1 in 200**.
- $P(\text{both parents transmitting a mutant allele})$: Finally, assuming both parents are carriers, the probability of both parents independently transmitting a mutant allele is $(1/2) \times (1/2)$, or **1/4**.

$$P(\text{affected child}) = 2/3 \times 1/200 \times 1/4 = 1 \text{ in } 1,200$$

Therefore, the probability of this couple conceiving an affected child is 1 in 1,200.

Educational objective:



AA

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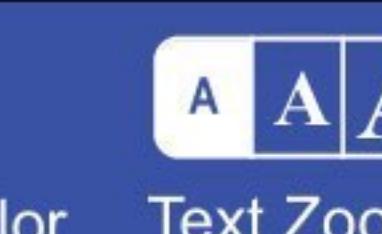
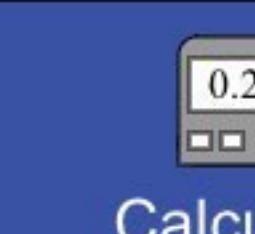
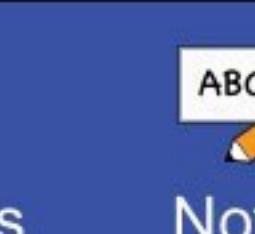
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following genotypes in the woman:

- Homozygous mutant
- Heterozygous carrier with her mother's mutant allele
- Heterozygous carrier with the father's mutant allele
- Homozygous normal

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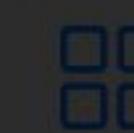
- P(carrier father): The man's genotype is unknown and he has **no relevant family history**; therefore, the probability of him being a carrier is equal to the carrier frequency in the general population, or **1 in 200**.
- P(both parents transmitting a mutant allele): Finally, assuming both parents are carriers, the probability of both parents independently transmitting a mutant allele is $(1/2) \times (1/2)$, or **1/4**.

$$P(\text{affected child}) = 2/3 \times 1/200 \times 1/4 = 1 \text{ in } 1,200$$

Therefore, the probability of this couple conceiving an affected child is 1 in 1,200.

Educational objective:

The probability that two healthy parents will conceive a child with an autosomal recessive disorder is equal to the probability of both parents being carriers multiplied by the probability of the child inheriting a mutant allele from each parent.



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Text Zoom

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Molecular biologists undertake a series of experiments designed to classify proteins involved in various intracellular signaling pathways. During one of the experiments, a protein mixture obtained from a cell culture is separated by gel electrophoresis and subsequently transferred to a filter membrane. Labeled double-stranded DNA probes are then used to detect a specific protein of interest in the sample. Which of the following proteins is most likely to be detected by this method?

- A. Ras (35%)
- B. c-Jun (27%)
- C. β 1-adrenoreceptor (3%)
- D. S-100 (13%)
- E. Adenylate cyclase (19%)

Omitted

Correct answer

B



27%

Answered correctly



07 secs

Time Spent



2023

Version

Explanation

Blotting technique	Substance detected	Type of probe
Northern	RNA	Single-stranded DNA
Southern	DNA	or RNA

Blotting technique	Substance detected	Type of probe
Northern	RNA	Single-stranded DNA or RNA (hybridization probe)
Southern	DNA	
Western	Protein	Antibody
Southwestern	DNA-binding protein	Double-stranded DNA

The Southern, Western, Northern, and Southwestern blot procedures are powerful techniques used to analyze and identify DNA fragments, proteins, mRNA, and DNA-bound proteins, respectively. The same basic technique underlies all of the blot procedures. First, the unknown sample is separated by gel electrophoresis. Separation occurs based on a molecule's size and charge. The separated molecules form bands on the gel that are then blotted onto a nitrocellulose membrane and incubated with a labeled probe to identify the specific DNA fragment, RNA molecule, or protein of interest.

Southwestern blots are used to identify and isolate proteins that bind DNA. In this technique, the target protein binds to a labeled, double-stranded DNA probe that is homologous to the protein's regulatory sequence. Of the molecules listed, c-Jun is the only DNA-binding protein. c-Jun and c-Fos are nuclear transcription factors that directly bind DNA via a leucine zipper motif. The genes that code for c-Jun and c-Fos are proto-oncogenes, genes that can become oncogenes following a mutation or with constitutive expression.

(Choice A) Ras is a proto-oncogene that codes for a membrane-bound G-protein. This G-protein acts as a secondary mediator for several hormones and cytokines that act on cell membran



AA



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A A A



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(Choice A) Ras is a proto-oncogene that codes for a membrane-bound G-protein. This G-protein acts as a secondary mediator for several hormones and cytokines that act on cell membrane receptors. Ras activation activates the MAP kinase pathway and ultimately affects transcription. However, Ras itself does not bind directly to DNA.

(Choices C and E) The $\beta 1$ -adrenergic receptor is a classic G_s -protein-coupled receptor located in the cell membrane. It does not interact directly with DNA. Adenylyl cyclase is the enzyme that cleaves ATP to form cAMP, the second messenger associated with G_s -protein-coupled receptors. cAMP activates protein kinase A for further downstream signaling.

(Choice D) S-100 proteins are homodimeric calcium-binding proteins, similar in structure to calmodulin and important in intracellular functions such as protein phosphorylation and cell growth and differentiation. S-100 is a marker for cells of neural crest derivation (melanocytes and Schwann cells), as well as Langerhans cells and other dendritic cells.

Educational objective:

Southwestern blotting is used to detect DNA-binding proteins such as transcription factors, nucleases, and histones.

References

- [Southwestern blotting in investigating transcriptional regulation.](#)