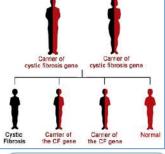
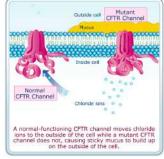
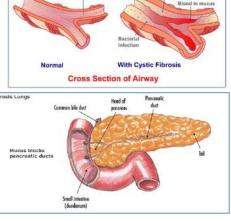
Chapter II – Human Genetic Disorders – CP Biology	
I. Major types of genetic disorders:	
•	
o	
o	
•	
•	
2. Autosomal genetic disorders are caused by	
Most are (need 2 recessive alleles to have the disorder the disorder content of the disorder determined of the disord	er)
 People with 1 recessive allele are they do NOT 	
are able to	
• Ex: cystic fibrosis (CF), sickle-cell anemia	
Can also be (need only I allele to have the disorder))
• Ex: Huntington's disease	
A) Cystic Fibrosis	6 4
CF is the most common genetic disorder among	
\circ ~I in 2500 white infants in the US are born with CF (4-5 born each day)	Carrier of Carr cystic fibrosis gene cystic fibr
o It is estimated that I in 20 white people is a carrier of the CF allele	1 1 1
Caused by an abnormal gene on	TTT
 The gene is for a that uses 	Cystic Carrier of Carrier o Fibrosis the CF gene the CF ge
to regulate the movement of sodium (Na*) and	Outside cell CF
chloride (Cl ⁻) ions into and out of cells	
 In healthy individuals, the normal protein 	Inside cell
	Normal CFTR Channel
 Keeps mucus thin and 	Chloride ians o
 With CF, not enough Cl⁻ ions are pumped out of cells 	A normal-functioning CFTR channel ions to the outside of the cell while channel does not, causing sticky mu on the outside of the ce
 in airways & pancreation 	tic ducts
Symptoms of CF:	Airway wall Thin layer of mucus Thick mucus
 Buildup of mucus in 	
Difficulty	Bacterial
•	Normal With Cystic Fib
 Blocks (produced by the rosis Lungs) 	Cross Section of Airway
	Head of Panarealic Common taile duct panareas duct

• Abnormal Na⁺ transport also results in _____







•	Treatments	for	CF:
---	------------	-----	-----

•	
•	
•	
• in severe cases	
 For digestive symptoms: 	
Capsules containing	
B) Sickle-Cell Anemia ()	
The most common genetic disorder among	
 About I in 500 African Americans has sickle-cell anemia. 	
 Carriers are said to have sickle-cell 	
Caused by an abnormal gene on	
 The gene is for one of the polypeptide chains in, a 	
protein found in that is responsible for	Typical Sickle Cell Trait Sickle Cell Disease
	(No Blood Disorder)
 Sickle-cell anemia causes hemoglobin to within red blood 	🍣 🏠 😫
cells, from the normal	
biconcave disc to a sickle shape.	NORMAL CLUMPED HEMOGLOBIN HEMOGLOBIN
 People with sickle-cell trait have some 	HEMOGLOBIN HEMOGLOBIN
but do not have the symptoms of sickle-cell disease	Sickle cell
Symptoms of Sickle-Cell Anemia:	Normal red
 Abnormal hemoglobinas 	blood cell
efficiently to cells as in healthy individuals	
•	
•	
•	
 Sickled red blood cells cannot move as easily through 	as normal RBCs
 Chronic, especially in 	
•to infections	
•	Sickle Cells
Treatments for Sickle-Cell Anemia:	Normal Red Blood Cells
o	
o	Sickle cells blocking blood flow
 that increase the oxygen-carrying capacity of red blood cells 	
 Drugs that "switch on" the gene for hemoglobin (normally sw 	itched off after birth)

• Heter	ozygote Superiority		
0	Sickle-cell anemia is most common in a	areas of the world where	is prevalent
	 Malaria is caused by a parasite 	that	
	 These parasites do not thrive 	in people with	, so
	people with sickle-cell trait are	e to mal	aria
0	People who are heterozygous for the	cystic fibrosis allele may be m	nore resistant to
0	When have an adva	ntage over people who are _	,
	it is called		
C) Hunting	ton's Disease	●┬□	
Caused	l by an		
(unlike	most human genetic disorders)	• 0 t	
0	Both men & women		to get the disorder
• Sympto	oms of Huntington's disease		
0	Huntington's disease affects a person's	i	Huntington's Disease Affects the Brain's Basal Ganglia
	•		(alles
	•		
	•		Basal
	•		Ganglia
	 Loss of muscle coordination a 	nd ability to speak	
0	Symptoms normally appear by		
0	Huntington's disease is always		
	 Death normally occurs within 	after t	he onset of symptoms
3. Many genetic	c disorders are believed to be the result	t of	:
•			
•			
•			
0	Bipolar disorder, schizophrenia		
These	are much more complicated to analyze	than disorders caused by sing	gle genes
4. Sex-linked di	isorders are almost always caused by m	utant alleles on the	
•			Inheritance of Red-Green Color Blindness: an X-linked Recessive Trait
•			
0	Women can be,	, but men cannot	
0	Ex: Homozygous normal female: X ^B X ^B Carrier female: X ^B X ^b		

= Carrier of Trait

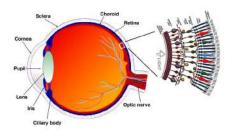
Carrier female:

XbXb XBY Colorblind female: Normal male: XbY

Colorblind male:

A) Hemophilia is caused by an abnormal gene for _____

- Blood does not clot normally, so even a tiny cut can result in ______
- _____ is also a major concern
 - Most common around ______
- Hemophiliacs ______
- B) Red-green colorblindness is caused by an abnormal gene for
 - The genes for both red and green photoreceptors are located on the X chromosome colorblindness can result from recessive alleles for either one or both of these genes



5. Chromosome abnormalities are caused by mistakes made during meiosis

- May change the ______ or _____ of chromosomes in the gametes that are formed
 - ______ the failure of a pair of chromosomes

to separate during meiosis

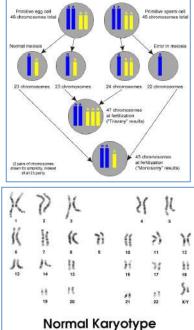
- Results in one gamete having too many chromosomes and another too few
- a zygote gets 3 copies of a chromosome
- _____ a zygote gets only I copy of a chromosome
- ________ is when a piece of one chromosome
 breaks off and attaches to a different chromosome
 - Often happens to 2 chromosomes at once
- Both nondisjunction and translocation can be detected in _____
 - Made from taking individual pictures of all of a human's chromosomes and matching up _____

A) **Down syndrome** – a genetic disorder that results from chromosome abnormality

- Nondisjunction the person has an extra copy of
- Translocation most of chromosome 21 breaks off during meiosis and fuses with another chromosome
 - Symptoms of Down syndrome:
 - Mild to severe ______
 - _____
 - · ____

Susceptibility to _____

and



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Section 2

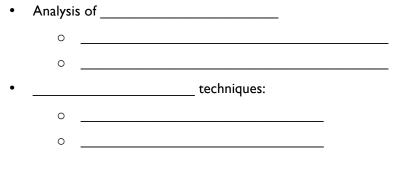
I. _____ disabilities are different from genetic disorders

 Occur during ______

2. Both genetic and congenital disorders can often be detected _____

- 3. Genetic _____
 - Can help parents determine the ______ of their child being born with a genetic disorder
 - Genetic counselors study the _____ of both parents
 - Create _______ to trace the passage of traits
 - _____ analyze blood tests to determine if parents are of certain genetic disorders
 - Usually can NOT determine whether or not a child will be born with a disorder, only the probability

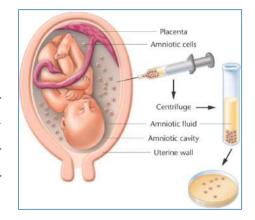
4. Two main ways to diagnose genetic disorders:



A) Amniocentesis

- _____ is the fluid that surrounds a fetus inside the uterus
 - Also contains fetal cells
- A sample of amniotic fluid is taken and cells are grown in a lab
 - Can be used to make a ______
 - Can be analyzed for ______
 - Detects ______

Cannot be conducted until ______



B) Chorionic Villus Biopsy

between a mother and developing fetus (they are part of the) • The villi develop from and therefore contain the				
 developing fetus (they are part of the) The villi develop from and therefore contain the same as the fetus & anniotic fluid A sample of these cells can be taken and analyzed as in amniocentesis 	•		•	
 The vill develop from and therefore contain the same as the fetus & anniotic fluid A sample of these cells can be taken and analyzed as in anniocentesis 				
<pre>same as the fetus & amniotic fluid . A sample of these cells can be taken and analyzed as in amniocentesis</pre>				Yolk sac
 A sample of these cells can be taken and analyzed as in amniocentesis 	•	The villi develop from	and therefore contain the contain th	ne Alexandre
 A sample of these cells can be taken and analyzed as in amniocentesis 		same	as the fetus & amniotic fluid	Embr
 Can be done as early as	•	A sample of these cells can be taken	and analyzed as in amniocentesis	
 Can be done as early as		0		
C) Ultrasonography • Uses high-frequency which bounce off of tissue • Depending on the of the tissue, the waves "echo" back at different and are used to produce a computerized image called an • Used in most pregnancies to detect • Used with amniocentesis to • Used with amniocentesis to • Can also help doctors detect abnormalities such as • D) Fetoscopy • A is made in a pregnant woman's • An on the end that on a monitor • Has a on the end that on a monitor • can be inserted through the endoscope tube to S. Developing cures for genetic disorders: A) Gene therapy •		0		ingun de ban fann e
 Uses high-frequency which bounce off of tissue Depending on the of the tissue, the waves "echo" back at different Introducing pregnancies to detect	•	Can be done as early as		
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 Depending on the of the tissue, the waves "echo" back at different and are used to produce a computerized image called an Used in most pregnancies to detect Used with amniocentesis to Can also help doctors detect abnormalities such as Can also help doctors detect abnormalities such as D) Fetoscopy A	C) L	Jltrasonography		
 Depending on the of the tissue, the waves "echo" back at different and are used to produce a computerized image called an Used in most pregnancies to detect Used with amniocentesis to Can also help doctors detect abnormalities such as Can also help doctors detect abnormalities such as D) Fetoscopy A	•	Uses high-frequency	which bounce off	of tissue
 Used in most pregnancies to detect				
 Used with amniocentesis to		and are	e used to produce a computerized i	mage called an
 Used with amniocentesis to	•	Used in most pregnancies to detect		
 Can also help doctors detect abnormalities such as	•			
D) Fetoscopy A				
 A		•		
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 An	•		is made in a pregnant woman's	
 Has a on the end that on a monitor can be inserted through the endoscope tube to 5. Developing cures for genetic disorders: A) Gene therapy Introducing into the cells of people with 0 0<!--</td--><td>•</td><td></td><td></td><td></td>	•			
 can be inserted through the endoscope tube to	•			
 5. Developing cures for genetic disorders: A) Gene therapy Introducing into the cells of people with Using				
A) Gene therapy Introducing into the cells of people with Using Using alleles in, which are taken into the cell by Currently these are still 		o C	an de inserted through the endosco	pe tube to
A) Gene therapy Introducing into the cells of people with Using Using alleles in, which are taken into the cell by Currently these are still 				
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 Using	A) (Г	/
 Using	•	Introducing	into the cells of people with	DNA encoding Factor VIII
 Enclosing alleles in, which are taken into the cell by Currently these are still 			-	Factor VIII Proteins
 Enclosing alleles in, which are taken into the cell by Currently these are still 		 Using 		王雪邁
 are taken into the cell by Currently these are still 				Factor VIII Gene
are taken into the cell by Currently these are still		• Enclosing alleles in	, which	
Currently these are still				Nucleus
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				Homan Cent