GENETIC DISORDER RESEARCH POWER POINT – PREZI– ANIMOTO PROJECT

A Collaborative Research Project Biology Adapted from Claire Carey Santa Barbara High School UCSB RET II 2006

CP BIO

Genetics Disorder Research Project

This unit project focuses on how genetics affects humans directly. Many people are affected by genetic disorders; you may even have some family members or friends with some of these diseases. I suggest that you choose a disorder that affects someone you know or intrigues you because it is strange or interesting. The goal is to research the disorder and orally present information and recent research through a group PowerPoint.

Genetic Disorder	Description	Websites
Alzheimer's Disease	A progressive brain disorder that gradually destroys a person's memory and ability to learn, reason, make judgments, and communicate.	http://www.ygyh.org/?syndrome http://www.alz.org http://www.alzheimers.org http://www.alzheimers.org.uk http://www.nlm.nih.gov/medlineplus/alzheimersdise ase http://www.namashealth.com/Alzheimers http://www.alzforum.org http://www.alzinfo.org/
Angelman Syndrome/ Prader-Willi Syndrome	Prader-Willi syndrome is an uncommon inherited disorder characterized by mental retardation, decreased muscle tone, and life- threatening obesity. When this genetic mutation is inherited from the mother, Angelman Syndrome arises which causes neurological problems including jerky movements and spontaneous laughter.	http://www.ncbi.nlm.nih.gov/books/bv.fcgi?rid=gnd.section.235http://www.ninds.nih.gov/disorders/angelman/angelmanhttp://ghr.nlm.nih.gov/condition=angelmansyndromehttp://www.angelman.orghttp://www.pwsausa.orghttp://www.pwsresearch.org/default.htmlhttp://www.ncbi.nlm.nih.gov/books/bv.fcgi?call=bv.ViewShowSection&rid=gnd.section.165

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Celiac Disease (<i>Celiac</i> <i>Sprue</i>)	A disease that triggers an autoimmune response that causes damage to the small intestine when certain types of protein, called gluten, are eaten.	http://www.nlm.nih.gov/medlineplus/co http://www.csaceliacs.org/celiac http://digestive.niddk.nih.gov/ddisease http://www.gluten.net/celiac http://www.labtestsonline.org/understa es/celiac http://www.celiac.org	s/pubs/celiac
Cri du chat Syndrome (<i>Cat's Cry</i> <i>Syndrome,</i> <i>Monosomy</i> <i>5p</i>)	Cri du chat syndrome is a group of symptoms that result from missing a piece of <u>chromosome</u> number 5. The syndrome's name is based on the infant's cry, which is high pitched and sounds like a cat.	http://www.icomm.ca/geneinfo/criducha http://ghr.nlm.nih.gov/condition=criduc http://gslc.genetics.utah.edu/units/diso pe/criduchat.cfm http://www.madisonsfoundation.org http://www.ncbi.nlm.nih.gov/entrez/qu http://members.tripod.com/%7Eyenial/	<u>chatsyndrome</u> <u>rders/karyoty</u> <u>ery</u>
Cystic Fibrosis	A recessive genetic disease in which the exocrine glands of afflicted individuals produce abnormally thick mucus that block the intestines and lung passageways. People with the disease have a very hard time breathing and often die from suffocation.	www.ygyh.org/?syndrome http://www.cff.org/home http://www.ncbi.nlm.nih.gov/books/bv.t ewShowSection&rid=gnd.section.242 http://www.cfri.org/home http://www.nlm.nih.gov/medlineplus/en 0107.htm http://learnaboutcf.tripod.com http://www.cysticfibrosis.com	
Diabetes, type 1 (Juvenile Diabetes) and Type 2	Diabetes is a chronic metabolic disorder that adversely affects the body's ability to manufacture and use insulin, a hormone necessary for the conversion of food	http://www.ncbi.nlm.nih.gov/books/bv.t ection.229 http://diabetes.niddk.nih.gov/dm/pubs/ x.htm http://familydoctor.org/480.xml http://www.jdrf.org	

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	into energy.	http://www.diabetes.org/home.jsp	
		http://ndep.nih.gov	
Down	Down syndrome is a chromosome	www.ygyh.org/?syndrome	
Syndrome	abnormality, usually	http://www.ndss.org	
(Trisomy 21)	due to an extra copy of the 21st chromosome. This syndrome	http://www.nads.org	
	usually, although not always, results in	http://www.down-syndrome.info	
	<u>mental retardation</u> and other conditions.	http://www3.ncbi.nlm.nih.gov/entrez/dis d=190685	<u>spomim.cgi?i</u>
		http://www.nas.com/downsyn/benke.htm	<u>nl</u>
Duchenne &	The muscular dystrophies are a	www.ygyh.org/?syndrome	
Becker Muscular	group of genetic diseases characterized	http://www.ncbi.nlm.nih.gov/disease/DM	<u>ID.html</u>
Dystrophy	by progressive weakness and	http://www.mdausa.org	
	degeneration of the skeletal muscles that	http://www.mayoclinic.com/invoke.cfm?	id=DS00200
	control movement.	http://www.nlm.nih.gov/medlineplus/mu phy.html	<u>usculardystro</u>
		<u>http://www.parentprojectmd.org/news/latml</u>	atest/index.h
Fragile X	Fragile X is a hereditary/genetic	www.ygyh.org/?syndrome	
Syndrome	condition caused by a mutation on the X	www.fragilex.org	
	chromosome. It can	http://www.fraxa.org	
	cause learning disabilities, or severe intellectual	http://www.ncbi.nlm.nih.gov/disease/FM	IR1.html
	complications including autism.	http://www.nlm.nih.gov/medlineplus/en- 1668.htm	<u>cy/article/00</u>
		http://www.nfxf.org/html	
Hemophilia	Hemophilia is a rare inherited bleeding	www.ygyh.org/?syndrome	
A or B	disorder that causes blood problems in	http://www.hemophilia.org/home	
	blood clotting.	http://www.wfh.org	
		http://www.ncbi.nlm.nih.gov/disease/her	moA.html
		www.hemophiliagalaxy.com	

		http://www.nlm.nih.gov/medlineplus/hemophilia.ht ml
		http://www.ncbi.nlm.nih.gov/books/bv.fcgi?rid=gnd.s ection.95
Huntington	A hereditary,	www.hdsa.org
Disease	degenerative brain disorder for which	www.ygyh.org/?syndrome
	there is no effective treatment or cure. HD	http://www.hdac.org
	slowly diminishes the affected individual's ability to walk, think, talk and reason.	<u>http://www.ncbi.nlm.nih.gov/disease/Huntington.ht</u> <u>ml</u>
	taik and reason.	http://www.huntington-assoc.com
		http://www.hdfoundation.org
		http://hdlighthouse.org
Klinefelter	In addition to	http://www.aaa.dk/TURNER/ENGELSK/INDEX.HT
Syndrome	occasional breast	M
CXXY	enlargement, lack of	
Syndrome)	facial and body hair, and a rounded body	http://www.nichd.nih.gov/publications/pubs/klinefelt er.htm
Syndronnel	type, XXY males are	
	more likely than other	http://www.genetic.org/ks/scvs/47xxy.htm
	males to be overweight, and tend	http://www.nlm.nih.gov/medlineplus/klinefelterssyn
	to be taller than their fathers and brothers.	<u>drome.html</u>
	fathers and provincis.	http://ghr.nlm.nih.gov/condition=klinefeltersyndrom
		<u>e</u>
		<u>http://www.ksa-uk.co.uk/</u>
		http://47xxy.org/
Maple Syrup Urine Disease	Maple Syrup Urine Disease is an	http://www.ncbi.nlm.nih.gov/books/bv.fcgi?rid=gnd.s ection.253
	inherited disorder so named because one of	http://www.meadjohnson.com/metabolics/maplesyru
	its first signs is urine	purinedesease.html
	that has an odor reminiscent of maple	http://www.msud-support.org/
	syrup.	
		http://www.nlm.nih.gov/medlineplus/ency/article/00 0373.htm
		http://www.savebabies.org/diseasedescriptions/msud .php

		http://rarediseases.about.com/od/rarediseases1/a/062 004.htm
Obesity	Obesity is an excess of body fat that	http://www.ncbi.nlm.nih.gov/books/bv.fcgi?rid=gnd.s ection.232
	frequently results in a significant impairment of health.	http://www.cdc.gov/genomics/info/perspectives/obesit y.htm
		http://www.nlm.nih.gov/medlineplus/obesity.html
		http://www.obesite.chaire.ulaval.ca/genes.html
		http://www.cancer.gov/newscenter/obesity1
		http://www.niddk.nih.gov/
Parkinson's Disease	Parkinson's disease is a motor system disorder which is the	http://www.ninds.nih.gov/disorders/parkinsons_dise ase/parkinsons_disease
result of dopamin brain cel Parkinso	result of the loss of dopamine-producing	http://www.apdaparkinson.org/user/index.asp
	brain cells. Parkinson's can cause	http://www.parkinson.org/site/pp.asp?c=9dJFJLPwB &b=71117
	tremors, rigidity, slowness of movement and postural	http://www.michaeljfox.org/
	instability.	http://www.parkinsonalliance.org/
		http://altrue.he.net/~thepi/site/parkinson/
Phenylketon	Phenylketonuria is a hereditary disorder in	www.ygyh.org/?syndrome
urią (<i>PKU</i>)	which the amino acid phenylalanine isn't	http://www.pkunews.org/
	properly metabolized. As a result, the amino acid can build up to	http://www.ncbi.nlm.nih.gov/disease/Phenylketo.ht ml
	dangerous levels in the blood and other	http://www.mayoclinic.com/invoke.cfm?id=DS00514
	tissues, causing mental retardation	http://www.pkunetwork.org
	and other serious health problems.	http://www.nlm.nih.gov/medlineplus/tutorials/pku
	neattii problems.	http://www.savebabies.org/diseasedescriptions/pku.p hp
		http://www.marchofdimes.com/professionals
Polycystic	Polycystic kidney disease is a genetic	www.ygyh.org/?syndrome

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Kidney Disease	disorder characterized by the growth of numerous cysts in the kidneys. The cysts can reduce kidney function and lead to kidney failure.	<u>http://kidney.niddk.</u> <u>ic</u> <u>http://familydoctor.o</u>	n.nih.gov/disease/PKD.html nih.gov/kudiseases/pubs/polycyst org/142.xml ov/condition=polycystickidneydis
Sickle Cell Anemia (<i>Sickle Cell</i> <i>Disease</i>) Tay-Sachs Disease	Sickle cell anemia is an inherited disorder that affects hemoglobin, a protein that enables red blood cells to carry oxygen to all parts of the body, resulting in a low number of red blood cells and periodic pain. A rare inherited disorder that causes progressive destruction of nerve cells in the brain and spinal cord, found to be more common in people of Ashkenazi Jewish heritage than in those with other backgrounds.	http://www.sicklecel http://www.sicklecel ml http://www.sicklecel ml http://www.ascaa.or http://ghr.nlm.nih.go http://ghr.nlm.nih.go http://www.marchof 27.asp http://www.genome. http://ghr.nlm.nih.go http://ghr.nlm.nih.go http://kidshealth.org counseling	<u>nih.gov/disease/sickle.html</u> <u>linfo.net</u> <u>ldisease.org/about_scd/faqs.pht</u> <u>g</u> <u>ov/ghr/disease/sicklecellanemia</u> <u>trome</u>
Triple-X Syndrome (<i>Trisomy X</i>)	A rare chromosomal genetic syndrome with one or more extra X chromosomes, leading to XXX (or more rarely XXXX or XXXXX), instead of the usual XX. These people are females	http://www.mayoclin 2&id=AN00351	CURNER/ENGELSK/INDEX nic.com/invoke.cfm?retryCount= ov/condition=triplexsyndrome sfoundation.org

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	and can be unaffected, or may suffer from problems such as infertility and reduced mental acuity.	http://www.med.umich.edu/1libr/yourcl http://www.wrongdiagnosis.com/t/triple /intro	-
Turner's Syndrome	Turner syndrome is a chromosomal condition that exclusively affects girls. It occurs when one of the two X chromosomes normally found in females is missing or incomplete.	http://www.aaa.dk/TURNER/ENGELS http://www.turner-syndrome-us.org/ http://www.tss.org.uk/ http://www.turnersyndrome.ca/ http://turners.nichd.nih.gov/ http://www.turnerssyndrome.org.au/	K/INDEX
XYY Syndrome	A condition in which males have two Y chromosomes; side effects include being more physically active and having a tendency to delayed mental maturation.	http://www.aaa.dk/TURNER/ENGELS http://www.ddhealthinfo.org/ggrc/doc2. =5199 http://my.webmd.com/hw/mens_conditi asp http://ghr.nlm.nih.gov/condition=47xyy http://www.nlm.nih.gov/medlineplus http://www.rarediseases.org	<u>asp?ParentID</u> ons/nord812.

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Group	Names:
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_____ Disorder: _____

Genetics Disorder Grading Rubric

Presentation Requirements	Earned Points	Possible Points	Comments
Introduction		20	
✓ Name the disorder			
✓ Introduce team members			
Definition of the disorder		20	
✓ What happens to the body?			
✓ What part of the body does it			
generally affect?		(2)	
Description of the symptoms		10	
✓ List all of the possible effects on			
the body			
Cause of the disorder		20	
✓ What happens <u>in the body</u> to			
cause the disease?			
✓ Is it a mutation? A genetic			
tendency triggered by other			
factors?		70	
How the disorder is inherited		30	
✓ Is it sex-linked?			
✓ Is there a particular chromosome			
it is located on?			
✓ Is it recessive or dominant?		10	
How the disorder is treated		10	
✓ Medications? Gene therapy?		10	
How the disorder is diagnosed		10	
✓ What tests are done? Is genetic			
counseling an option?		20	
How many and what type of people are likely to have the disorder		20	
,			
✓ Is it more common in a certain			
group of people? ✓ How common is it?			
Application of research article		10	
✓ What new information did you		10	
learn from your research?			
Total Points Earned		150	
		150	

Genetics Disorder Research Sheet

Research Site: _____

	Presentation Requirements	Research
Definitic	on of the disorder	
	What happens to the body?	
	What part of the body does it generally affect?	
Descripti	on of the symptoms	
✓	List all of the possible effects on the body	
	the disorder	
✓	What happens in the body to cause the disease?	
✓	Is it a mutation? A genetic tendency triggered by	
	other factors?	
How the	disorder is inherited	
✓	Is it sex-linked?	
✓	Is there a particular chromosome it is located on?	
✓	Is it recessive or dominant?	
How the	disorder is treated	
✓	Medications? Gene therapy?	
How the	disorder is diagnosed	
✓	What tests are done? Is genetic counseling an	
	option?	
How ma	ny and what type of people are likely to have the	
disorder		
✓	Is it more common in a certain group of people?	
✓	How common is it?	

Genetics Disorder Job Worksheet

Partner	Slides to Make & Present in Given Order		
	<u>Title Slide</u> : Project Title, team members		
	Definition Slide: What happens to body; part of body affected?		
	<u>Symptom Slide</u> : List of all effects on body		
	<u>Cause Slide</u> : What happens in body to cause disease; Mutation? Genetic tendency		
	triggered by other factors?		
	Inherited Slide: Sex-linked-yes or no; particular chromosome; recessive or		
	dominant?		
	<u>Treatment Slide</u> : Medications? Gene therapy?		
	Diagnostic Tests Slide: What tests are done; genetic counseling?		
	Frequency in Population Slide: More common in certain group; how common is		
	it?		
	Article Slide: New information found from research		

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Group	Names:	_
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_____ Disorder: _____

Genetics Disorder Self Evaluation

Presentation Requirements	Cumulative Research	
		Points
Definition of the disorder V What happens to the body? V What part of the body does it generally affect? Sites:		20
Description of the symptoms		
✓ List all of the possible effects on the body Sites:		10
Cause of the disorder		
 What happens in the body to cause the disease? Is it a mutation? A genetic tendency triggered by other factors? 		20
How the disorder is inherited		
 ✓ Is it sex-linked? ✓ Is there a particular chromosome it is located on? ✓ Is it recessive or dominant? 		30
Sites:		
How the disorder is treated ✓ Medications? Gene therapy? Sites:		10
How the disorder is diagnosed ✓ What tests are done? Is genetic counseling an option? Sites:		10
How many and what type of people are likely to have the disorder		
 Is it more common in a certain group of people? How common is it? 		20
Sites:		
Application of research article ✓ What new information did you learn from your research?		10
Resource:		
Total Points Earned		130

Genetic Disease: _____Group Names: _____Grader's Name: _____

Genetics Disorder Peer Review

Presentation Requirements	Earned Points	Possible Points	Comments
Introduction		20	
✓ Name the disorder			
 ✓ Introduce team members 			
Definition of the disorder		20	
 What happens to the body? 			
 What part of the body does it 			
generally affect?			
Description of the symptoms		10	
 ✓ List all of the possible effects on the 			
body			
Cause of the disorder		20	
 What happens <u>in the body</u> to cause 			
the disease?			
 Is it a mutation? A genetic tendency 			
triggered by other factors?			
How the disorder is inherited		30	
✓ Is it sex-linked?			
 Is there a particular chromosome it is 			
located on?			
✓ Is it recessive or dominant?			
How the disorder is treated		10	
✓ Medications? Gene therapy?			
How the disorder is diagnosed		10	
✓ What tests are done? Is genetic			
counseling an option?			
How many and what type of people are likely		20	
to have the disorder			
✓ Is it more common in a certain group			
of people?			
✓ How common is it?			
Application of research article		10	
 What new information did you learn 			
from your research?			
Total Points Earned		150	

What did you like about their presentation?

What could this group improve on?