**Joubert Syndrome**

Hailey Saunders, Brittany McNamara, and Sabrina Claeys



Biology Day 1 Block 2

10/25/13

**Joubert Syndrome**

1. **Title:**
	1. Living With Joubert
2. **Symptoms:**
	1. rapid breathing
	2. rapid, jerky eye movement
	3. inability to control muscle movements
	4. mental retardation
	5. extra toes or fingers
	6. cleft lip or palate
	7. coordination and balance problems
3. **Occurrence:**
	1. affects 1 in 80,000 to 1 in 100,000 newborns.
	2. More common in males. Easier to get in males.
		1. because of Y chromosome
	3. It is very rare because it affects less than 200,000 people in the US population
	4. More common in jewish families.
	5. More common in young children and through life.
4. **Inheritance:**
	1. has an autosomal recessive pattern of inheritance-both copies of a gene in each cell have mutations
	2. It affects at least 10 genes
	3. affects the X chromosome
	4. From the parents with joubert syndrome.
	5. Looked at by a punnett square
5. **Treatment:**
	1. symptomatic and supportive
	2. physical, occupational, and speech therapy may benefit some children
	3. infants with abnormal breathing should be monitored and if there’re any other problems with the body related to Joubert Syndrome, screenings should be done on a regular basis
	4. No cure.. Can have treatment for breathing
	5. For people with the severe disease may require more medical attention.
	6. Screening for progressive eye, liver, and kidney complications associated with Joubert-related disorders should be performed on a regular basis.
6. **Facts:**
	1. 3% of the US population has an intellectual disability.
	2. There are more than 4000 genetic disorders.
	3. 1 in 180 babies have an abnormal chromosome
	4. carry from 5 to 10 genes with mutations in each of their cells
	5. Genetic disorders are caused by too few or too much chromosomes or mutations.

**Log**

10/7: Chose Joubert and gathered information

10/9: Finished gathering information and Hailey began script

10/14: Everyone revised script and finished it and set days for recording

10/16: Got ¾ of recording done

10/20: Finished recording and Sabrina uploaded videos

10/21: Sabrina and Brittany began editing

10/22: Hailey made outline, and Brittany gathered music files

10/24: Uploaded the video and made sure everything was perfect. Finalized project to put on flashdrive

**Sources**

<http://ghr.nlm.nih.gov/condition/joubert-syndrome>

<http://www.babymed.com/genetics/joubert-syndrome>

<http://rarediseases.about.com/od/rarediseasesj/a/joubert.htm>

<http://www.patient.co.uk/doctor/Joubert-Syndrome.htm>

<http://www.chromosome18.org/AboutGenetics/FactsAboutGenetics/tabid/132/Default.aspx>

<http://www.rightdiagnosis.com/j/joubert_syndrome/prevalence.htm#prevalence_intro>

[http://kidshealth.org/teen/your\_body/health\_basics/genes\_genetic\_disorders.html#](http://kidshealth.org/teen/your_body/health_basics/genes_genetic_disorders.html)

Video Link

<http://www.youtube.com/watch?v=Hiz6GOYiwMk>