

Turner's Syndrome



A presentation by The Boss
Period 3

The background is a vibrant, abstract composition of overlapping organic shapes in various shades of pink, magenta, and lavender. The shapes are fluid and layered, creating a sense of depth and movement. The colors range from light, airy pinks to deeper, more saturated purples and magentas.

HI DR. NED

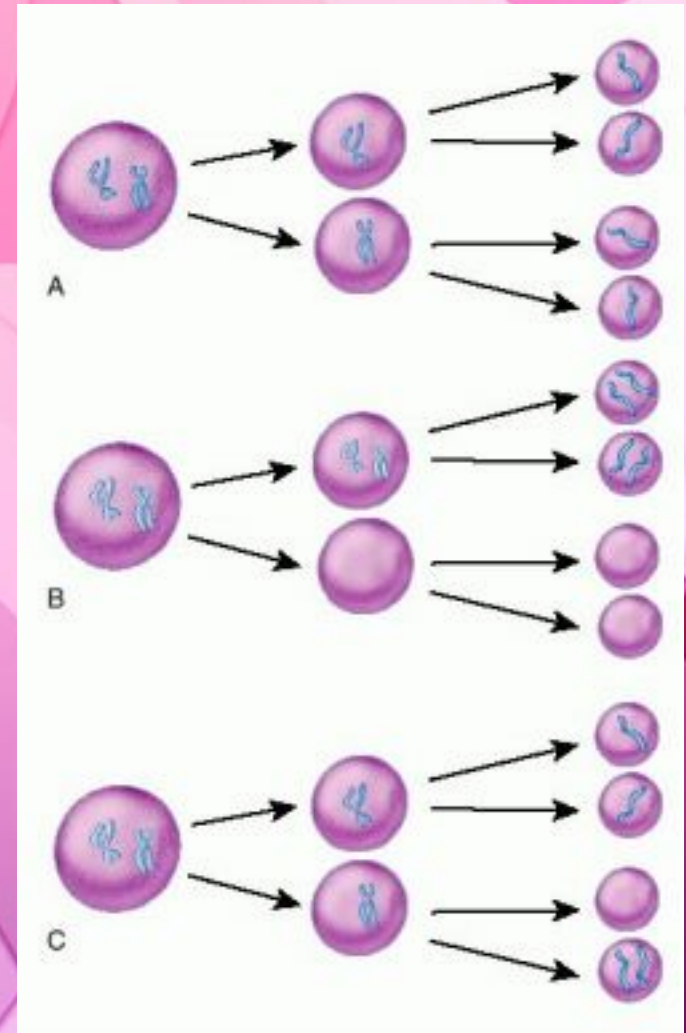
Physiology

- » Turner's syndrome occurs in approximately 1 per every 2000 babies.
- » Only females can be affected
- » Also known as Ullrich-Turner syndrome or Gonadal Dysgenesis
- » Age of onset is prenatal
- » Symptoms include but are not limited to: Swollen hands and feet, webbed necks, "shield chests", drooping eyelids, low set ears, receding jaws, and short stature
- » Lack of secondary sexual characteristics also such as: lack of pubic hairs, lack of curves, and absence of menstrual cycle
- » Increased risk of other health problems such as heart problems (usually in the left side of the heart), high blood pressure due to aorta and bicuspid aortic valve narrowing, hypothyroidism, ear infections, kidney problems, loss of hearing, cataracts, diabetes, arthritis, and scoliosis.



Molecular Cause/ Inheritance

- » Turner's syndrome is caused by a monosomy of the X chromosome. This means that there is only one X chromosome instead of two like normal females.
- » This is usually due to nondisjunction which occurs in meiosis. It can occur either during meiosis 1 or 2.
- » When it occurs in meiosis 1, two of the chromosomes don't separate and this causes for there to be two gametes with no genetic info and two with twice as much genetic info as they should have. In meiosis 2 it is possible for this to happen. The only difference is that it creates two normal gametes, 1 with no genetic info and 1 with twice of what it should have.



Treatments and Limitations

- » There is no cure for Turner's syndrome.
- » There are many therapies however which can help to improve the health problems of those with Turner's syndrome.
- » One therapy is estrogen replacement therapy. This therapy will help the affected female to develop secondary sexual characteristics. Limitation is that they have to keep going with this therapy until they reach menopausal age.
- » Another therapy is human growth hormone treatment. It helps them combat their short stature. The limitation is that it usually only increases the height by 8 cm at the most. They can also receive surgery for their neck if it is webbed.

Proposal

- » The reason why it is bad for females to have only one X chromosome is due to the X-inactivation system. This makes it so that every cell in a females body uses one X or the other. When someone has Turner's syndrome the X-inactivation knocks out the only X chromosome that they have.
- » There is a specific gene on every X-chromosome called the Xist gene which is in charge of the X-inactivation system. The Xist gene will inactivate the chromosome.
- » For my proposal, I have suggested that we use RNA interference to inhibit Xist gene expression. By doing this, since every cell only has one X-chromosome, if it is never inhibited then the affected female should be able to live a normal life.
- » Limitations to this is that RNAi is still being experimented with and it would be extremely difficult to target every Xist gene in the body.

Sources/References/Random Links

Title Page

http://eso-cdn.bestpractice.bmj.com/best-practice/images/bp/en-gb/1106-1-highlight_default.jpg

Physiology

"Turner Syndrome." *Turner Syndrome*. Ed. David Zieve. U.S. National Library of Medicine, 30 Mar. 2012. Web. 22 Feb. 2013.

Images

http://learn.genetics.utah.edu/content/disorders/whataregd/turner/images/turner_person.jpg

http://upload.wikimedia.org/wikipedia/commons/thumb/2/22/Neck_Turner.JPG/230px-Neck_Turner.JPG

Molecular Cause/Inheritance

<http://medical-dictionary.thefreedictionary.com/nondisjunction>

Treatments and Limitations

Genetic Science Learning Center. "Turner Syndrome." Learn.Genetics 21 February 2013
<learn.genetics.utah.edu/content/disorders/whataregd/turner/>

Proposal

"Intersection of the RNA Interference and X-Inactivation Pathways." *Sciencemag.org*. Sciencemag.org, 30 Apr. 2008. Web. 18 May 2013.