

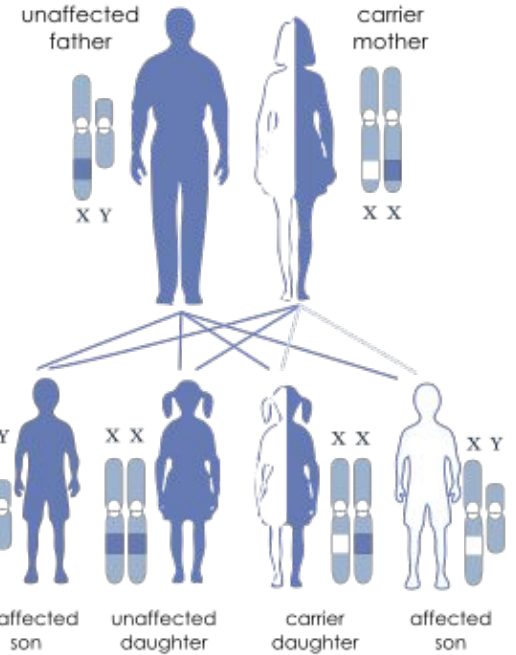
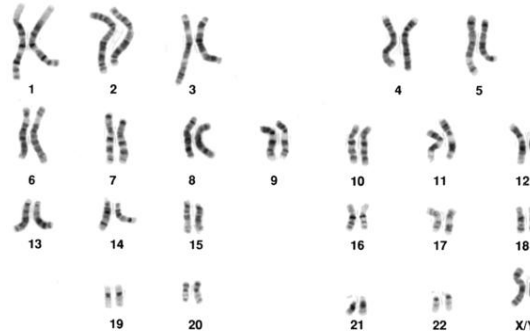
Arts Syndrome

By Emily



What chromosome/gene is affected?

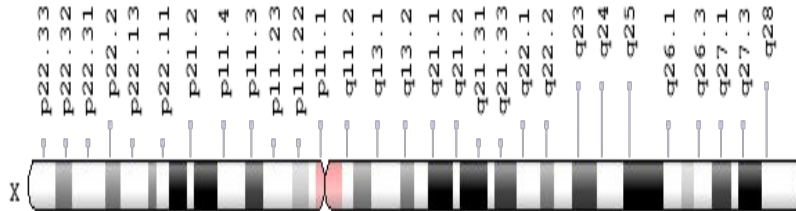
X-linked recessive inheritance



■ unaffected ■ carrier □ affected

The X chromosome is affected

Mutations in the PRPS1 gene



Normal Karyotype



Characteristics of disease

Boys

Cause serious neurological problems

Have extreme hearing loss from lesions or disease of inner ear or auditory nerve

Other characteristics include weak muscle tone, impaired muscle coordination, development delay, and intellectual disabilities

Girls

Milder symptoms

Lose their hearing when entering adulthood





Treatment

Cochlear implantation

Routine immunizations for common childhood infections and influenza

Regular neuropsychological and audiologic examinations

S-adenosylmethionine (SAME) which is a chemical compound found in most cells and tissues

Physical/ occupational therapy



Should we invest in research?

I think we should have more people look in to not just Arts Syndrome but other rare diseases. There are so many rare diseases out there that I think we should know a little more about them. Arts Syndrome wasn't discovered until 2007. If we do more research we might be able to find a cure to some of the diseases

But at the same time. Most are so rare only one or two people in the world has a rare disease. So should we invest money into looking more into a diseases if the majority of the population isn't affected?



How would my life change?

Life would change drastically if you had a child with Arts Syndrome. You would slowly start to see your son lose their hearing then their muscle tone. Your son may have some intellectual disabilities making learning for them hard. Then having to know that you won't be able to watch him grow up. Knowing that he'll die between the age of five and six. Then a miracle happens and your son lives past the life expectancy. You'll now need to get a wheelchair for him since their muscles are too weak to hold them. You'll also needed to get a respirator for him so they don't get lung infections and later on so he can breath. Your son will needed a special educational program due to his intellectual disability. Life will be hard. You'll needed to pay for his wheelchair and respirator. You'll also needed to pay for regular check ups.



How would my life change?Cont...

If I was informed that I was a carrier for this disease I would be worried that one of my future sons may get the disease. If I was pregnant I would want to check the gender of the baby as soon as I could. If it was a boy I would want to get a amniocentesis as soon as possible.

If the amniocentesis came back positive for Arts Syndrom I would be faced with a really hard decision. Should I abort the baby or have it? If I had it I'd have to be prepared for him to die in a few years. I think I would rather have the baby than never have it at all. Even if he's going to die at a young age you'll be able to have good memories about him, and you never know. You might get lucky and he'll live longer than expected.



Questions

Which chromosome is affected?

What kind of inheritance is it?

What's the life expectancy?

Why do you think this disease is so rare?



<https://ghr.nlm.nih.gov/condition/arts-syndrome#synonyms>

https://en.wikipedia.org/wiki/Arts_syndrome

<https://www.thinkgenetic.com/diseases/arts-syndrome/treatment/7195>