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# Heredity

## Mendelian ways of inheritance

**Véronique Nas**

4 October 2013

# Heredity

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## three ways of inheritance

**1** autosomal **dominant**

**2** autosomal **recessive**

**3** **x-linked**

autosomal =

not on the sex chromosomes

x-linked =

on the female sex chromosome

# Heredity

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**autosomal dominant: one mutated gene will do**

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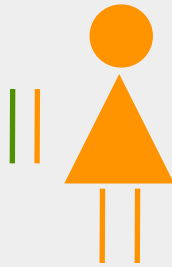
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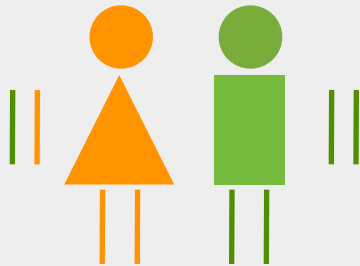


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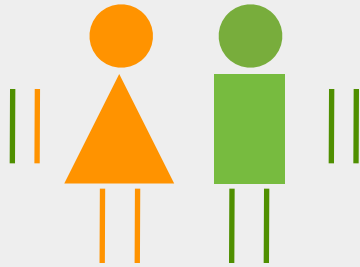


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## autosomal dominant: one mutated gene will do

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she marries a healthy man with two healthy genes  
if the woman passes on her mutated gene



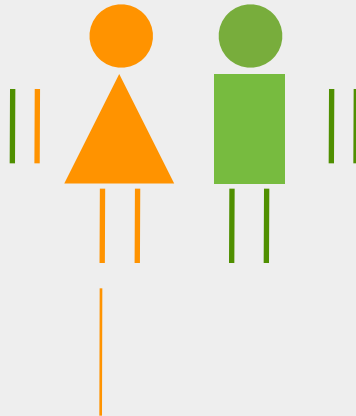


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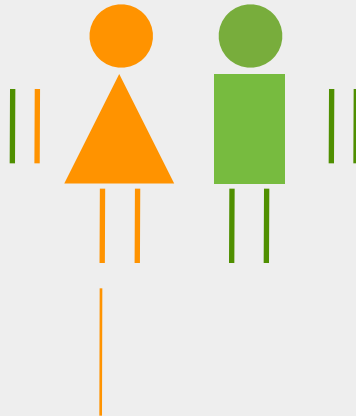


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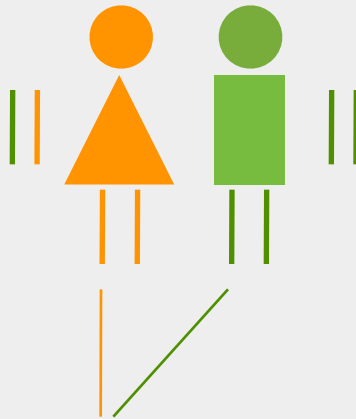


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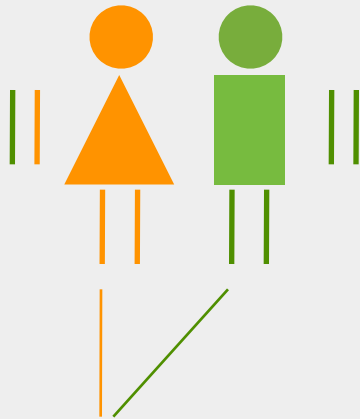
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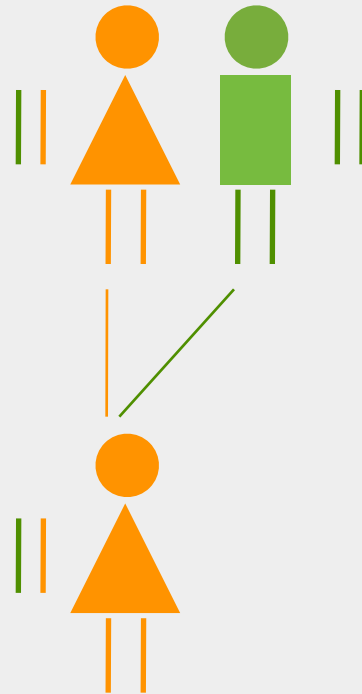
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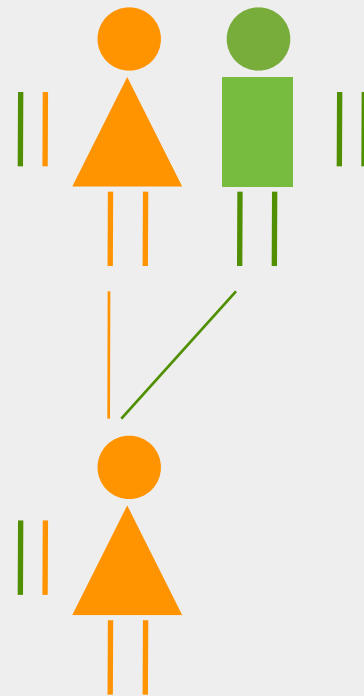
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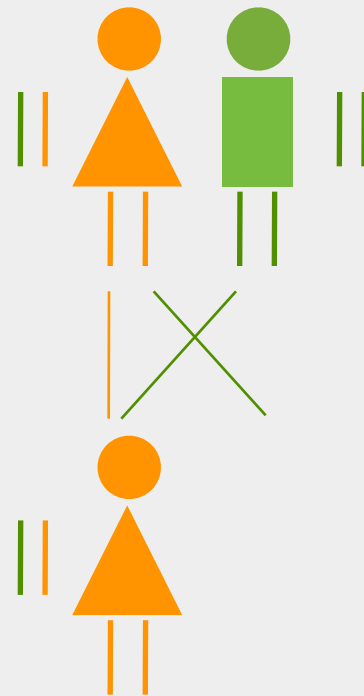
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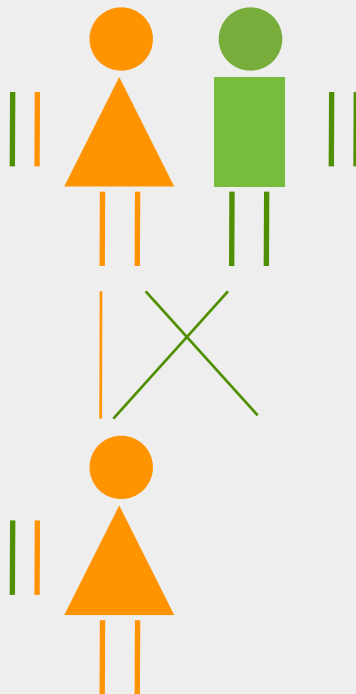
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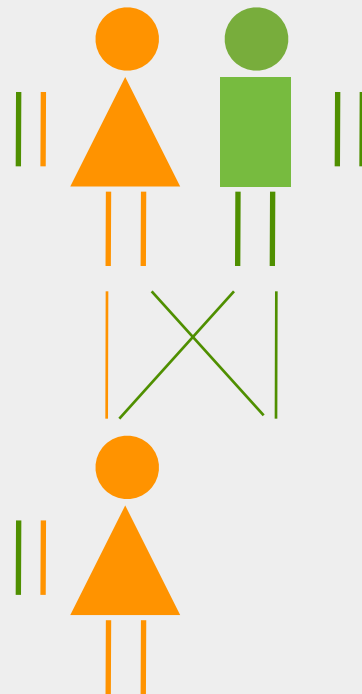
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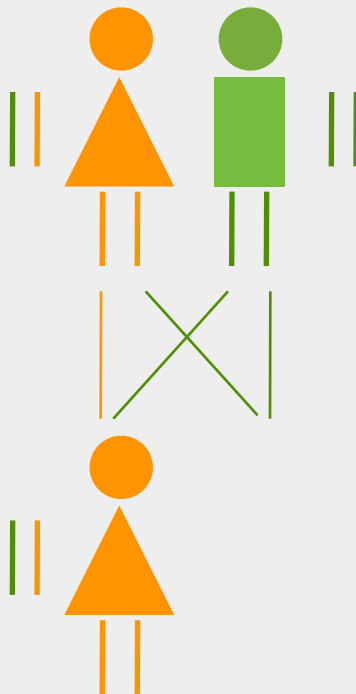
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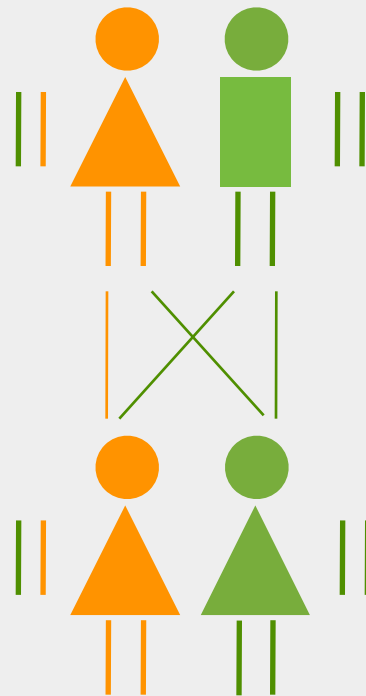
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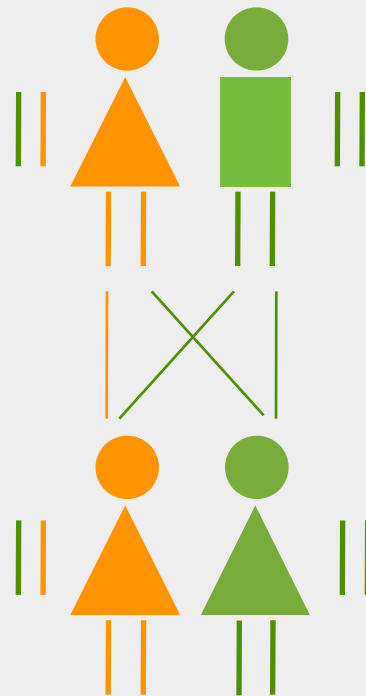
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whether it is a girl ...

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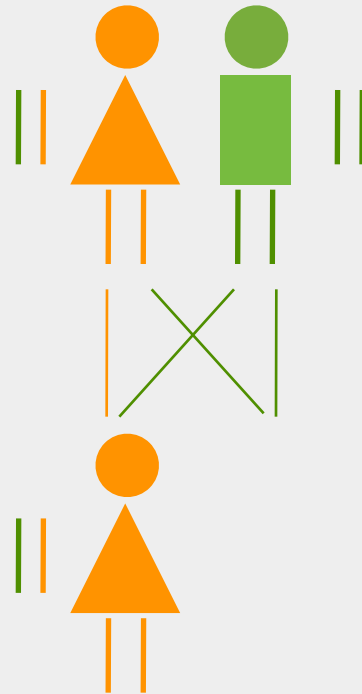
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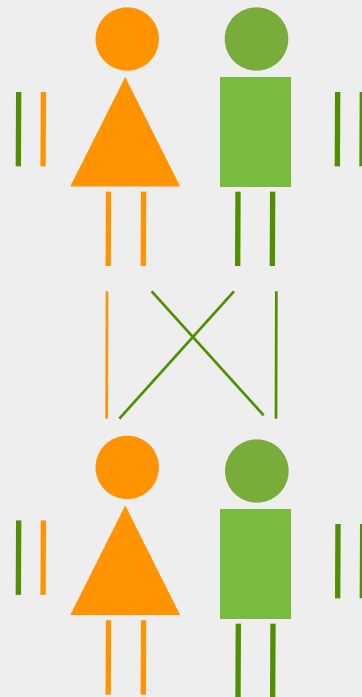
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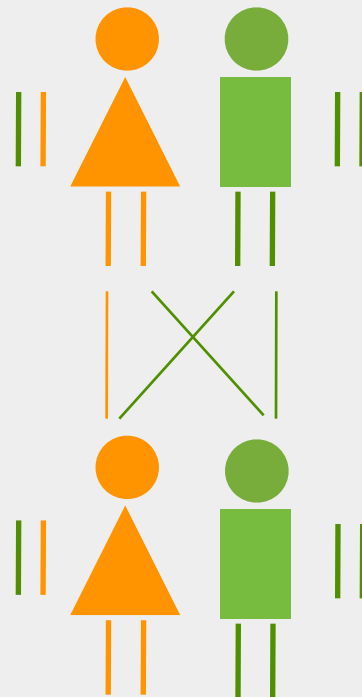
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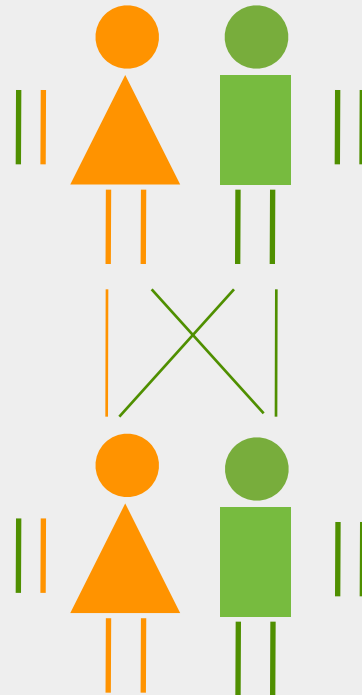
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every child has a 50% chance to inherit the disease



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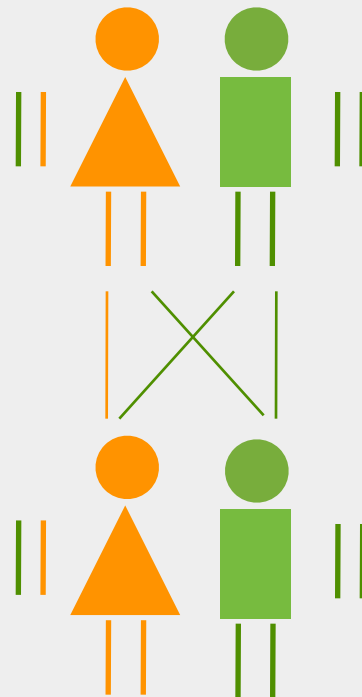
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an example is Wagner syndrome, a disease of the retina and vitreous, associated with many retinal detachments

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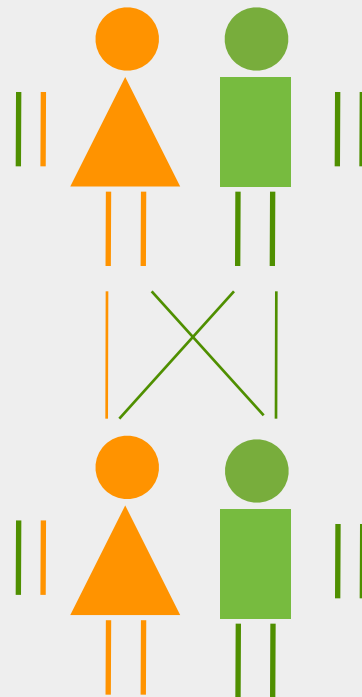
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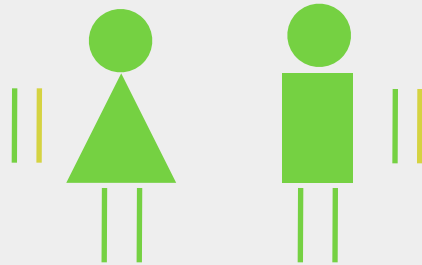
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**autosomal recessive: only 2 mutated genes cause the disease**

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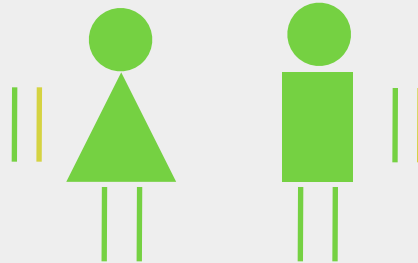


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**autosomal recessive: only 2 mutated genes cause the disease**

both parents have a healthy gene (green) and a mutated one (yellow)



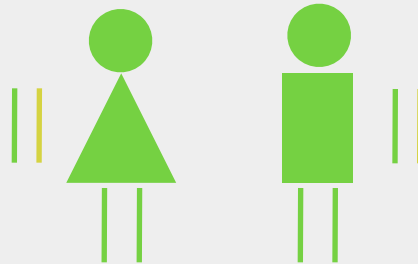
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both parents have a healthy gene (green) and a mutated one (yellow)

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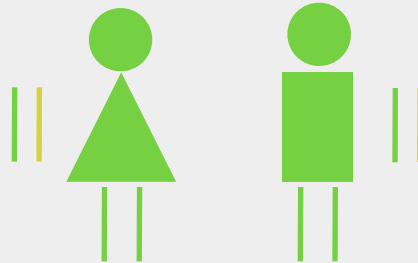
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## autosomal recessive: only 2 mutated genes cause the disease

both parents have a healthy gene (green) and a mutated one (yellow)

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only if both parents pass on the mutated gene to their child, it will inherit the disease



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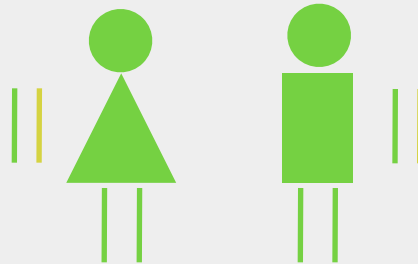
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there are four possibilities





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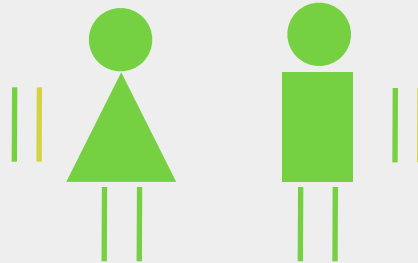
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woman healthy gene + man healthy gene = healthy child, no mutated genes

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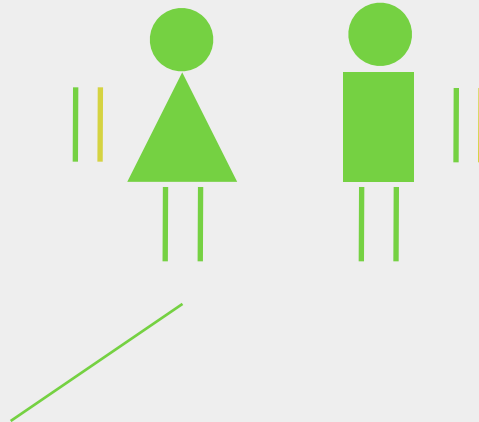
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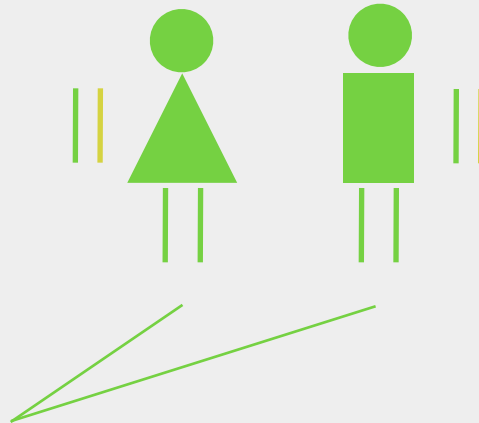
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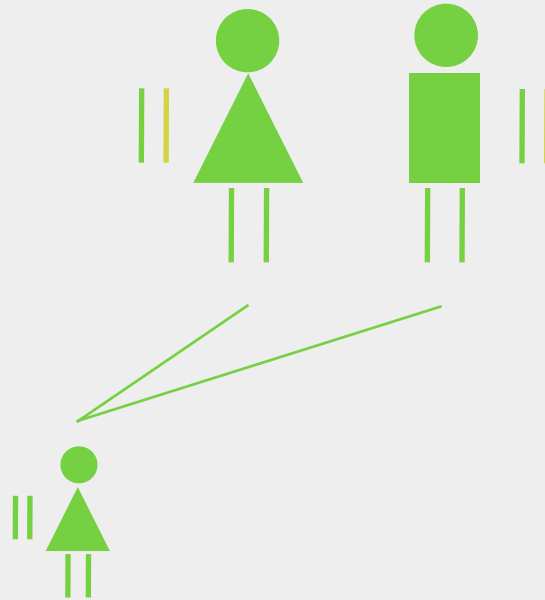
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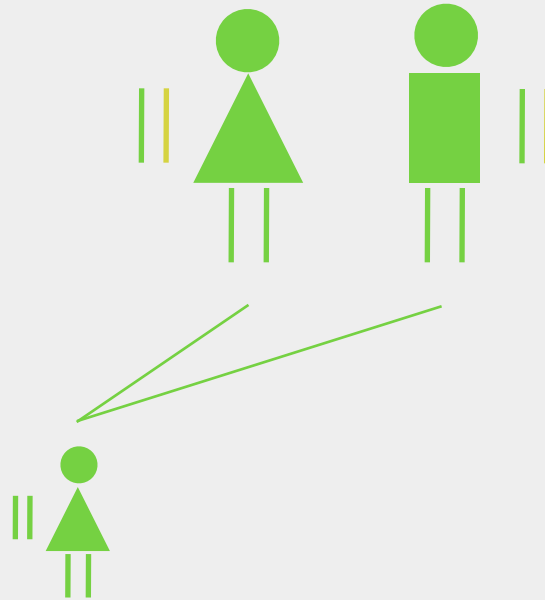
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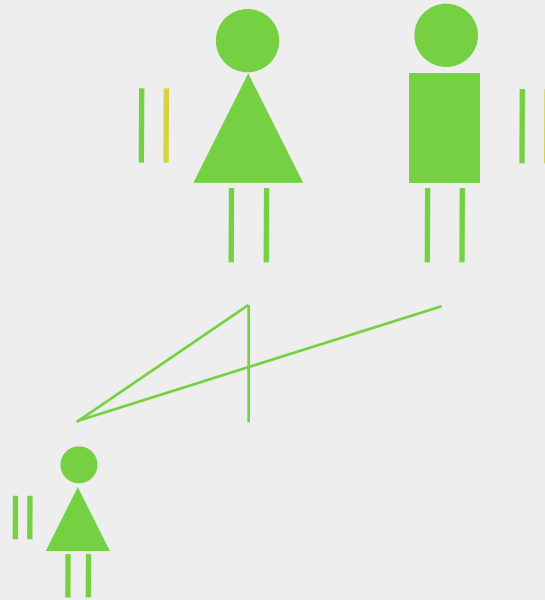
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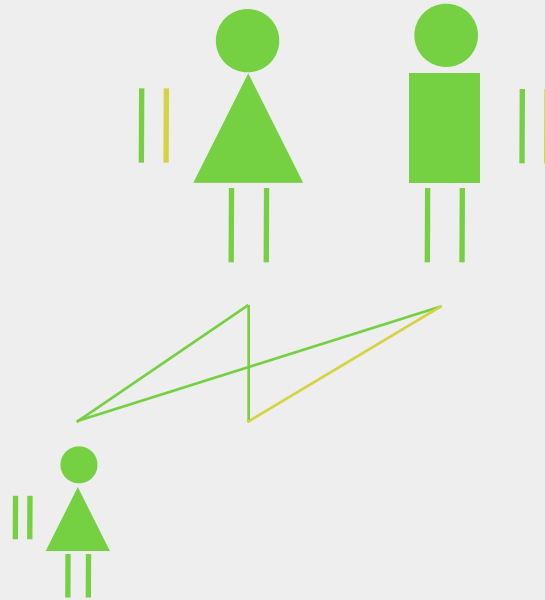
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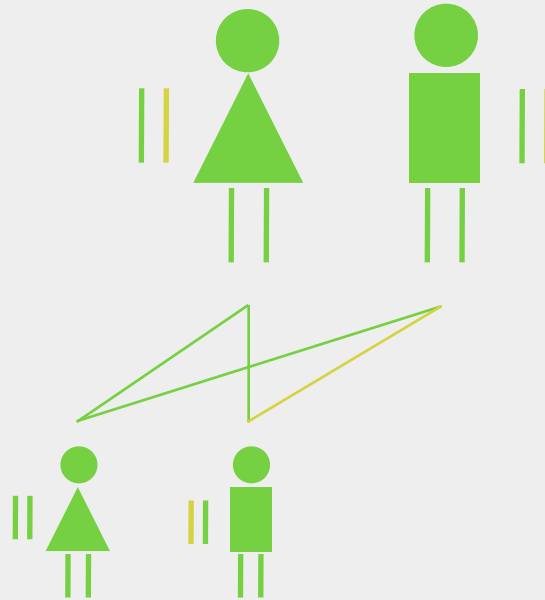
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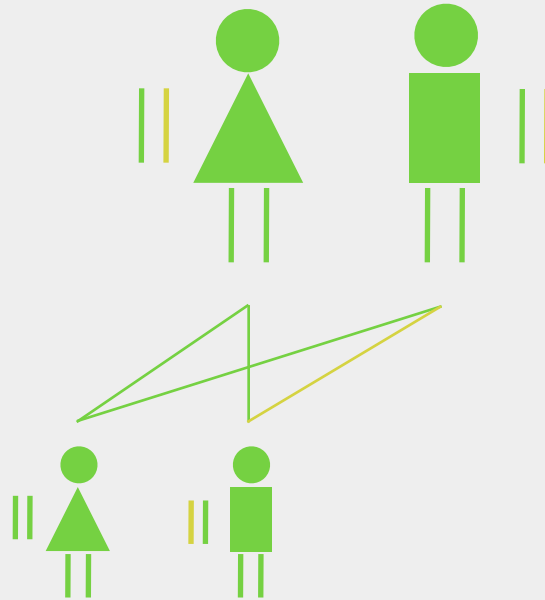
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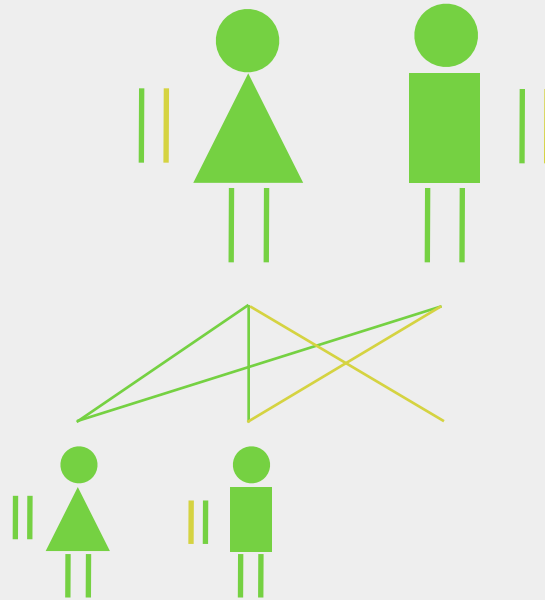
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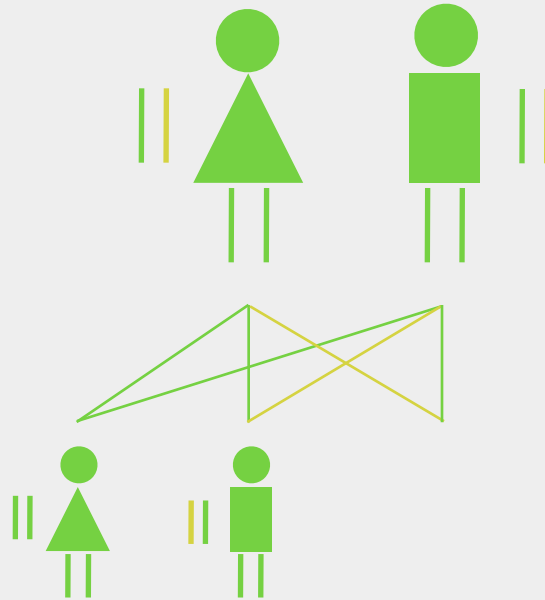
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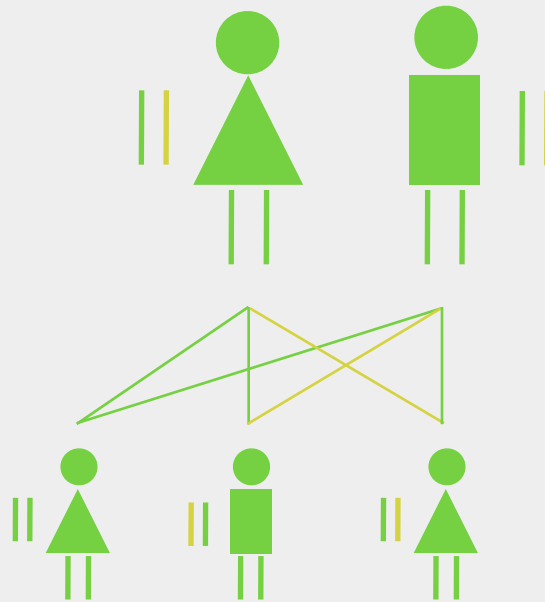
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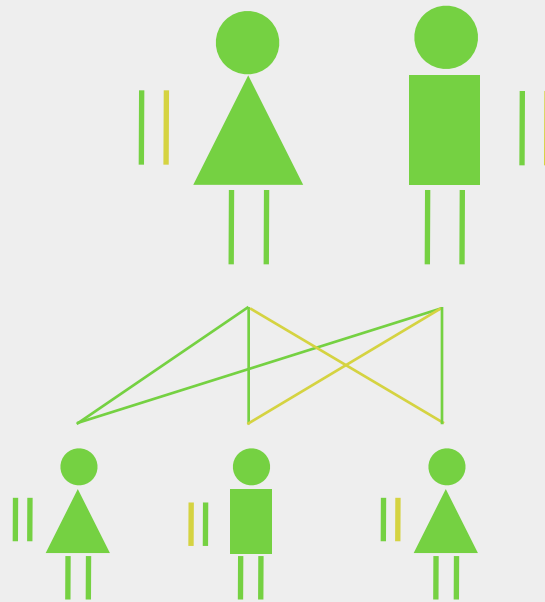
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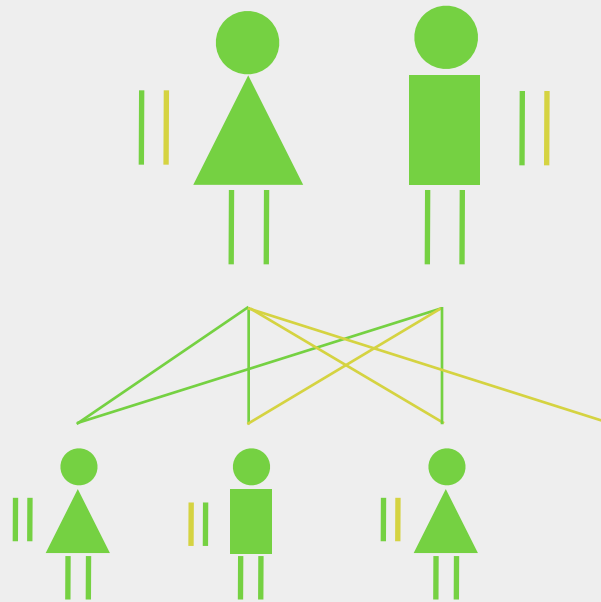
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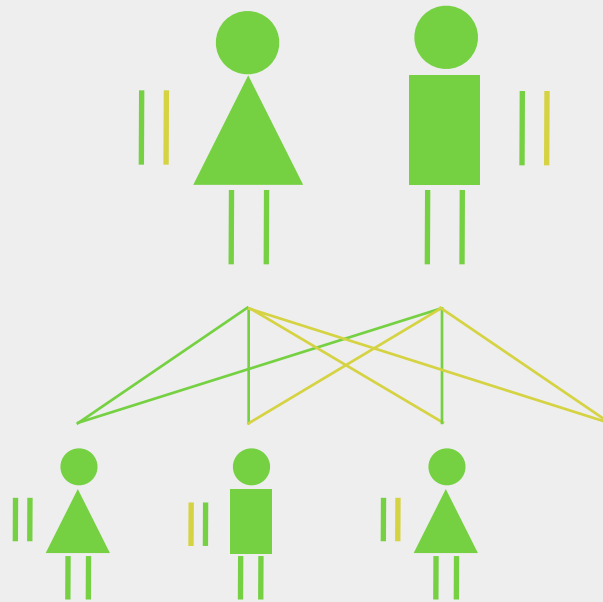
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woman sick gene + man sick gene  
= child with the disease, two sick genes

# Heredity

---

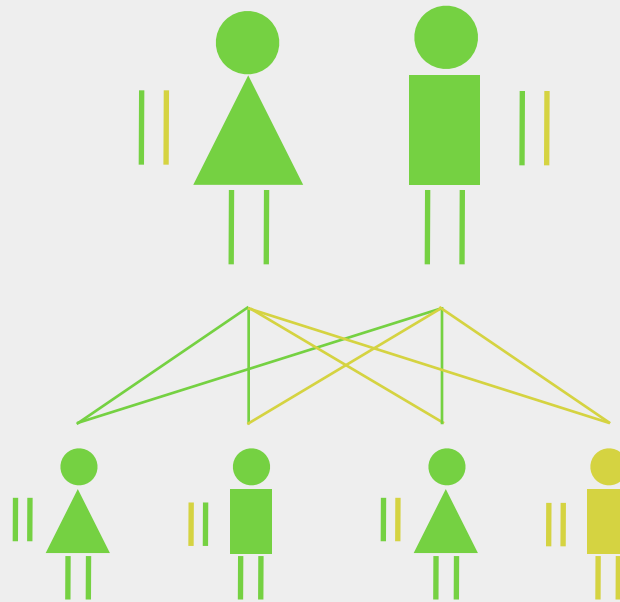
## autosomal recessive: only 2 mutated genes cause the disease

both parents have a healthy gene (green) and a mutated one (yellow)

as the healthy gene is dominant, neither of the parents carry the disease

only if both parents pass on the mutated gene to their child, it will inherit the disease

there are four possibilities



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# Heredity

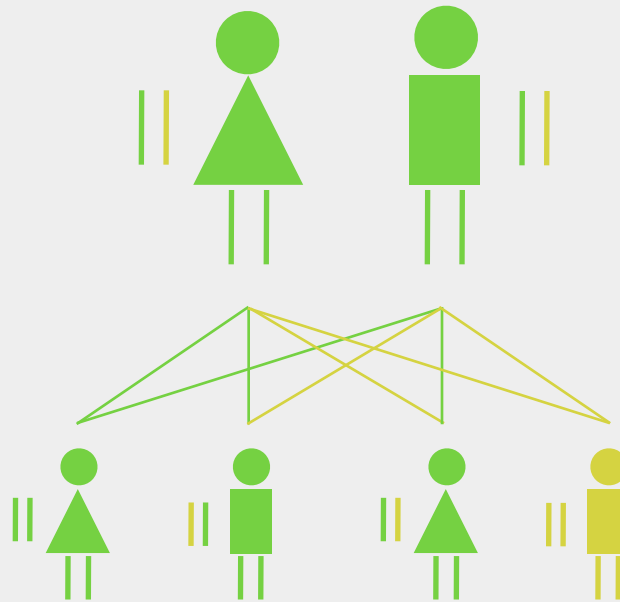
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= child with the disease, two sick genes

every child has a 25% chance of inheriting the disease

# Heredity

---

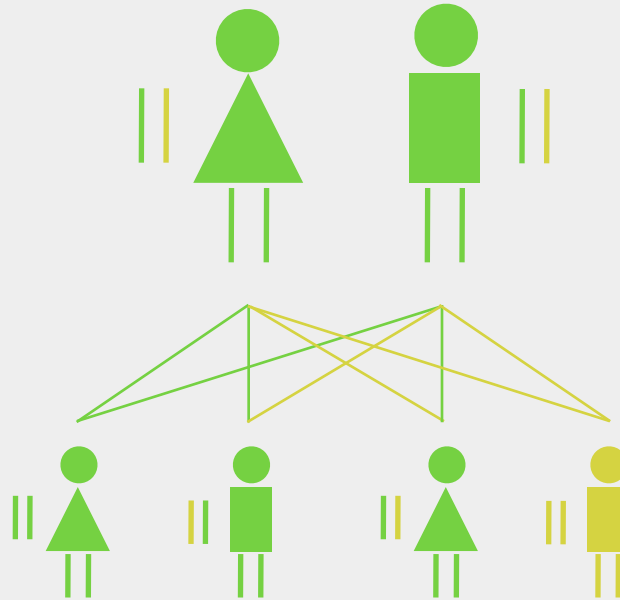
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= child with the disease, two sick genes

every child has a 25% chance of inheriting the disease

whether it is a girl or a boy

# Heredity

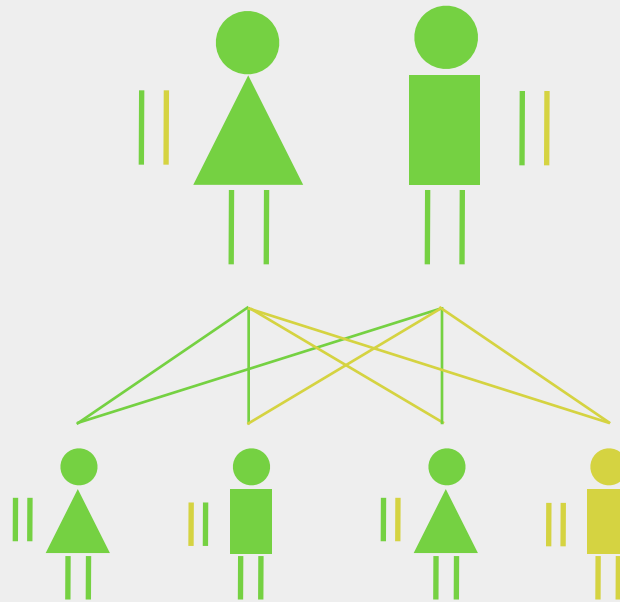
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every child has a 25% chance of inheriting the disease

whether it is a girl or a boy

75% of the children has a mutated gene.

they can only pass on the disease to their children if they marry a spouse with also a mutated gene

# Heredity

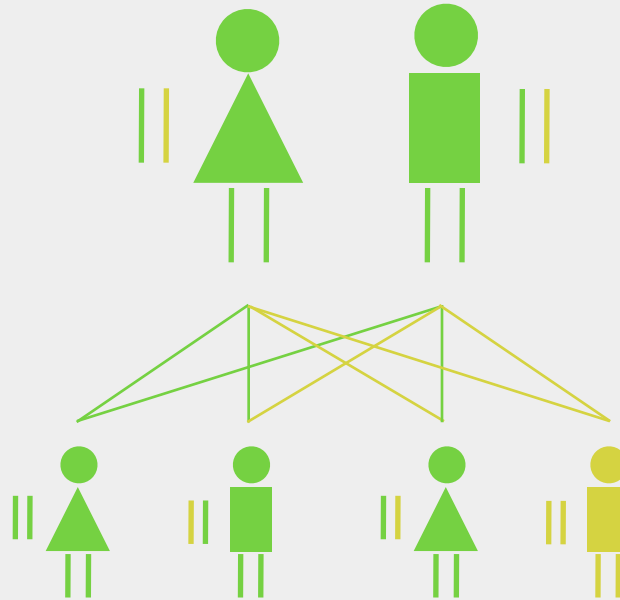
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every child has a 25% chance of inheriting the disease

whether it is a girl or a boy

75% of the children has a mutated gene.

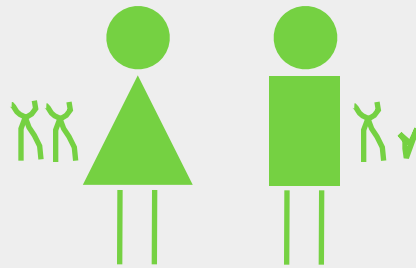
they can only pass on the disease to their children if they marry a spouse with also a mutated gene

an example is cystic fibrosis

# Heredity

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**x-linked: on the female sex chromosome, only boys at risk**

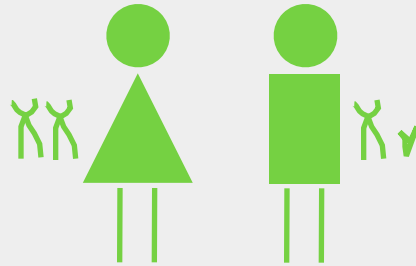


# Heredity

---

**x-linked: on the female sex chromosome, only boys at risk**

woman: two X-chromosomes



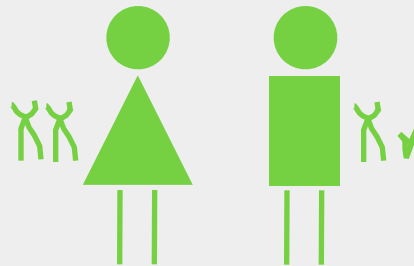
# Heredity

---

**x-linked: on the female sex chromosome, only boys at risk**

woman: two X-chromosomes

man: one Y-chromosome  
and one X-chromosome



# Heredity

---

## x-linked: on the female sex chromosome, only boys at risk

woman: two X-chromosomes

man: one Y-chromosome  
and one X-chromosome

on the (little) Y-chromosome  
there are almost only genes  
that determine the gender





# Heredity

---

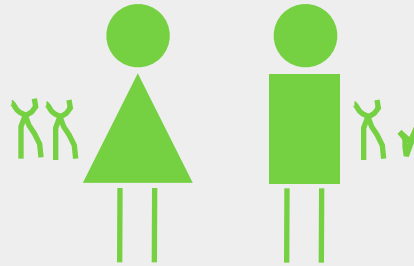
## x-linked: on the female sex chromosome, only boys at risk

woman: two X-chromosomes

man: one Y-chromosome  
and one X-chromosome

on the (little) Y-chromosome  
there are almost only genes  
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on the larger X-chromosome  
there are genes that  
determine the gender but  
also some other genes



# Heredity

---

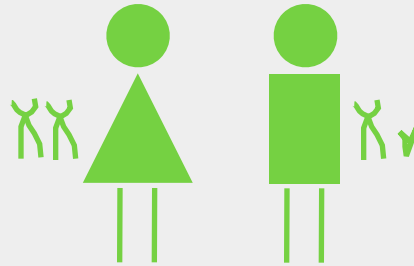
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also some other genes



let's say that on one of  
the X-chromosomes of  
a woman there is a  
mutated gene

# Heredity

---

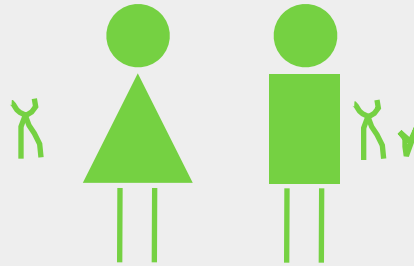
## x-linked: on the female sex chromosome, only boys at risk

woman: two X-chromosomes

man: one Y-chromosome  
and one X-chromosome

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there are genes that  
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let's say that on one of  
the X-chromosomes of  
a woman there is a  
mutated gene

# Heredity

---

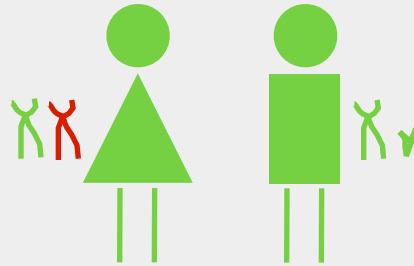
## x-linked: on the female sex chromosome, only boys at risk

woman: two X-chromosomes

man: one Y-chromosome  
and one X-chromosome

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let's say that on one of  
the X-chromosomes of  
a woman there is a  
mutated gene

# Heredity

---

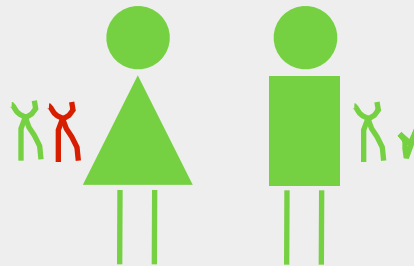
## x-linked: on the female sex chromosome, only boys at risk

woman: two X-chromosomes

man: one Y-chromosome  
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on the (little) Y-chromosome  
there are almost only genes  
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on the larger X-chromosome  
there are genes that  
determine the gender but  
also some other genes



let's say that on one of  
the X-chromosomes of  
a woman there is a  
mutated gene  
than we have four  
possibilities

# Heredity

---

## x-linked: on the female sex chromosome, only boys at risk

woman: two X-chromosomes

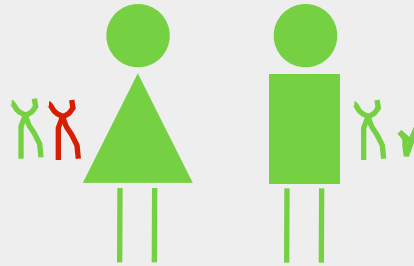
man: one Y-chromosome  
and one X-chromosome

on the (little) Y-chromosome

there are almost only genes  
that determine the gender

on the larger X-chromosome

there are genes that  
determine the gender but  
also some other genes



woman healthy X + man X-chromosome  
= healthy girl, no mutated genes

let's say that on one of  
the X-chromosomes of  
a woman there is a  
mutated gene  
than we have four  
possibilities

# Heredity

---

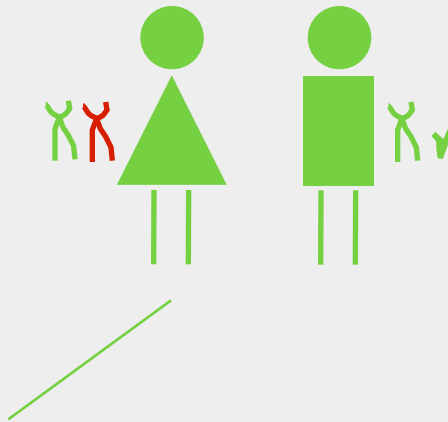
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woman healthy X + man X-chromosome  
= healthy girl, no mutated genes

let's say that on one of  
the X-chromosomes of  
a woman there is a  
mutated gene  
than we have four  
possibilities

# Heredity

---

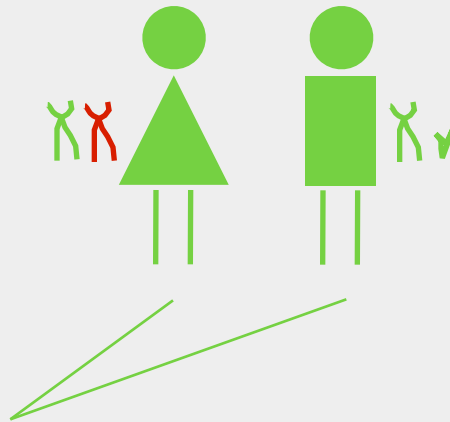
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= healthy girl, no mutated genes

let's say that on one of  
the X-chromosomes of  
a woman there is a  
mutated gene  
than we have four  
possibilities



# Heredity

---

## x-linked: on the female sex chromosome, only boys at risk

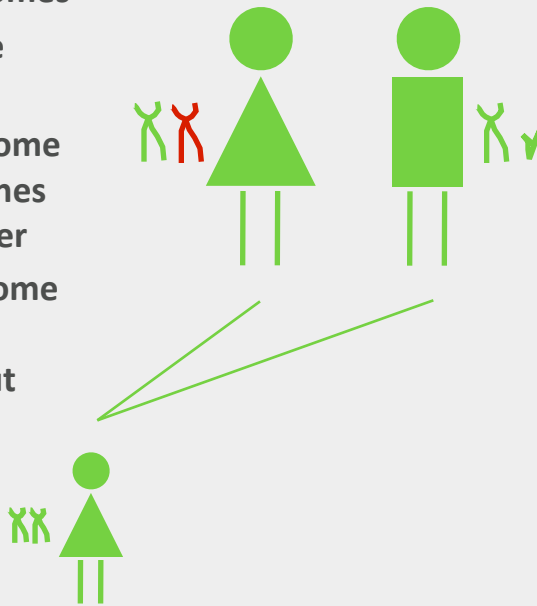
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there are almost only genes  
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also some other genes

let's say that on one of  
the X-chromosomes of  
a woman there is a  
mutated gene  
than we have four  
possibilities



woman healthy X + man X-chromosome  
= healthy girl, no mutated genes

# Heredity

---

## x-linked: on the female sex chromosome, only boys at risk

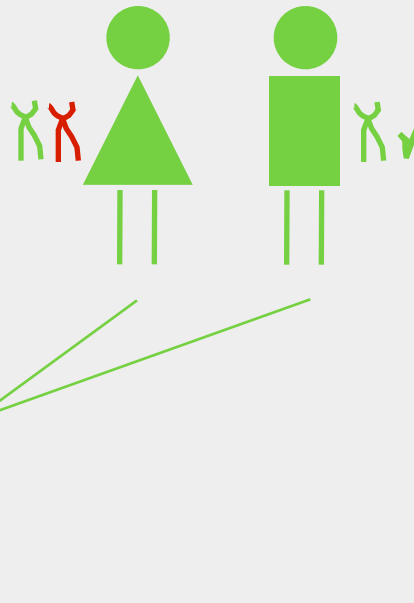
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on the (little) Y-chromosome  
there are almost only genes  
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there are genes that  
determine the gender but  
also some other genes

let's say that on one of  
the X-chromosomes of  
a woman there is a  
mutated gene  
than we have four  
possibilities



woman healthy X + man X-chromosome  
= healthy girl, no mutated genes

woman sick X + man X-chromosome  
= healthy girl, one mutated gene  
the healthy X makes the girl not having  
the disease



# Heredity

---

## x-linked: on the female sex chromosome, only boys at risk

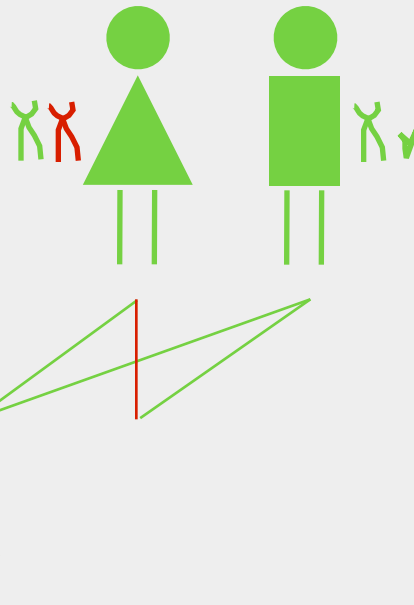
woman: two X-chromosomes

man: one Y-chromosome  
and one X-chromosome

on the (little) Y-chromosome  
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mutated gene  
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the healthy X makes the girl not having  
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# Heredity

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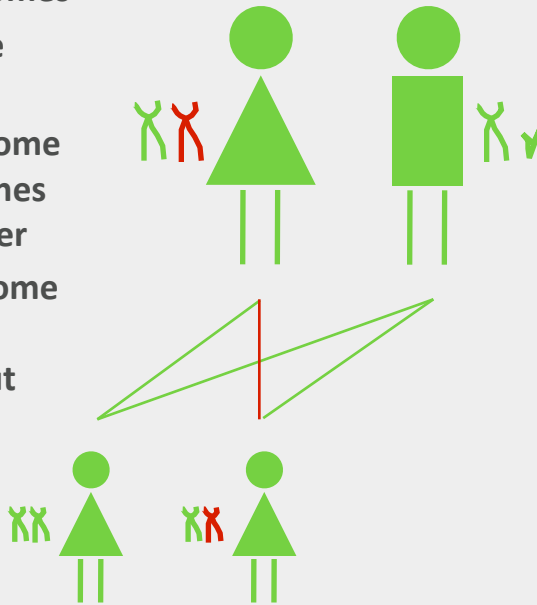
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man: one Y-chromosome  
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on the (little) Y-chromosome  
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determine the gender but  
also some other genes

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the X-chromosomes of  
a woman there is a  
mutated gene  
than we have four  
possibilities



woman healthy X + man X-chromosome  
= healthy girl, no mutated genes

woman sick X + man X-chromosome  
= healthy girl, one mutated gene  
the healthy X makes the girl not having  
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# Heredity

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## x-linked: on the female sex chromosome, only boys at risk

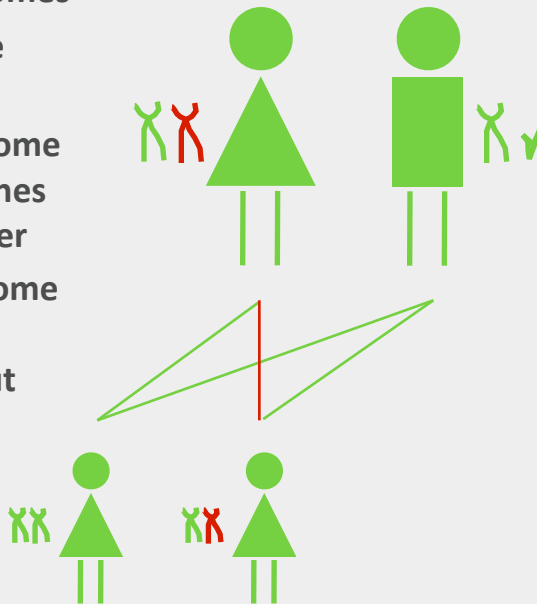
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man: one Y-chromosome  
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on the (little) Y-chromosome  
there are almost only genes  
that determine the gender

on the larger X-chromosome  
there are genes that  
determine the gender but  
also some other genes

let's say that on one of  
the X-chromosomes of  
a woman there is a  
mutated gene  
than we have four  
possibilities



woman healthy X + man X-chromosome  
= healthy girl, no mutated genes

woman sick X + man X-chromosome  
= healthy girl, one mutated gene  
the healthy X makes the girl not having  
the disease

woman healthy X + man Y-chromosome  
= healthy boy

# Heredity

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## x-linked: on the female sex chromosome, only boys at risk

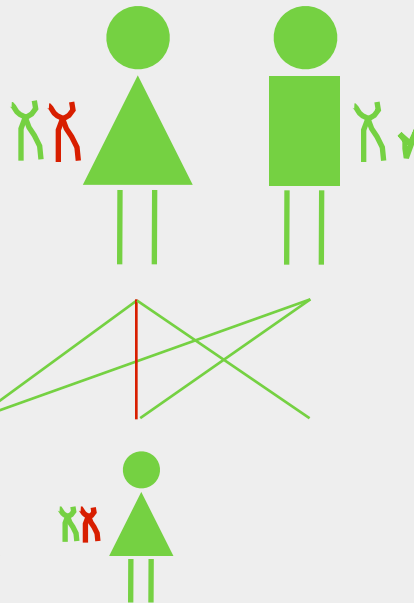
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that determine the gender

on the larger X-chromosome  
there are genes that  
determine the gender but  
also some other genes

let's say that on one of  
the X-chromosomes of  
a woman there is a  
mutated gene  
than we have four  
possibilities



woman healthy X + man X-chromosome  
= healthy girl, no mutated genes

woman sick X + man X-chromosome  
= healthy girl, one mutated gene  
the healthy X makes the girl not having  
the disease

woman healthy X + man Y-chromosome  
= healthy boy

# Heredity

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## x-linked: on the female sex chromosome, only boys at risk

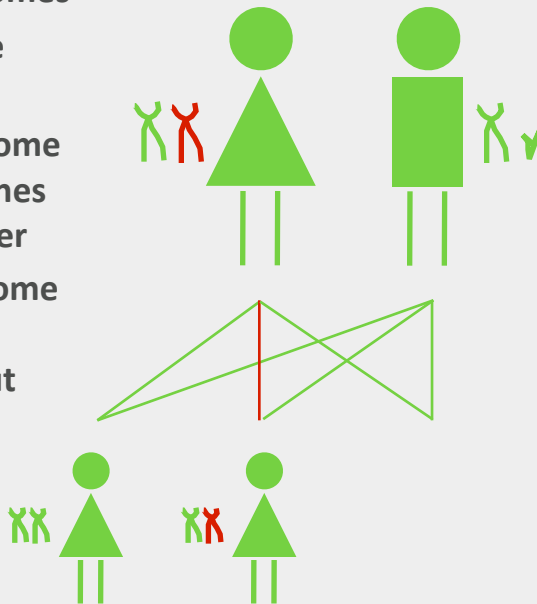
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man: one Y-chromosome  
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possibilities



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= healthy girl, one mutated gene  
the healthy X makes the girl not having  
the disease

woman healthy X + man Y-chromosome  
= healthy boy



# Heredity

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## x-linked: on the female sex chromosome, only boys at risk

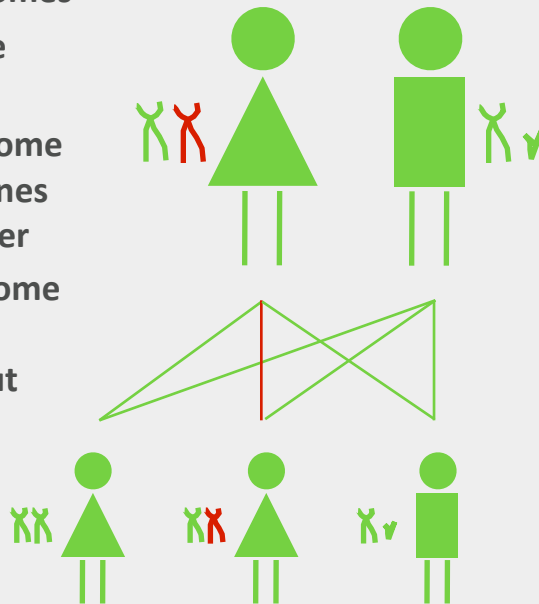
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mutated gene  
than we have four  
possibilities



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= healthy girl, no mutated genes

woman sick X + man X-chromosome  
= healthy girl, one mutated gene  
the healthy X makes the girl not having  
the disease

woman healthy X + man Y-chromosome  
= healthy boy

# Heredity

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## x-linked: on the female sex chromosome, only boys at risk

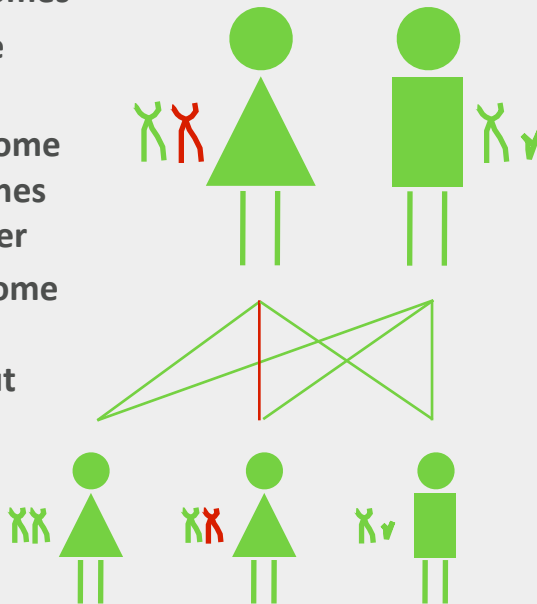
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on the (little) Y-chromosome  
there are almost only genes  
that determine the gender

on the larger X-chromosome  
there are genes that  
determine the gender but  
also some other genes

let's say that on one of  
the X-chromosomes of  
a woman there is a  
mutated gene  
than we have four  
possibilities



woman healthy X + man X-chromosome  
= healthy girl, no mutated genes

woman sick X + man X-chromosome  
= healthy girl, one mutated gene  
the healthy X makes the girl not having  
the disease

woman healthy X + man Y-chromosome  
= healthy boy

woman sick X + man Y-chromosome  
= boy with the disease

# Heredity

## x-linked: on the female sex chromosome, only boys at risk

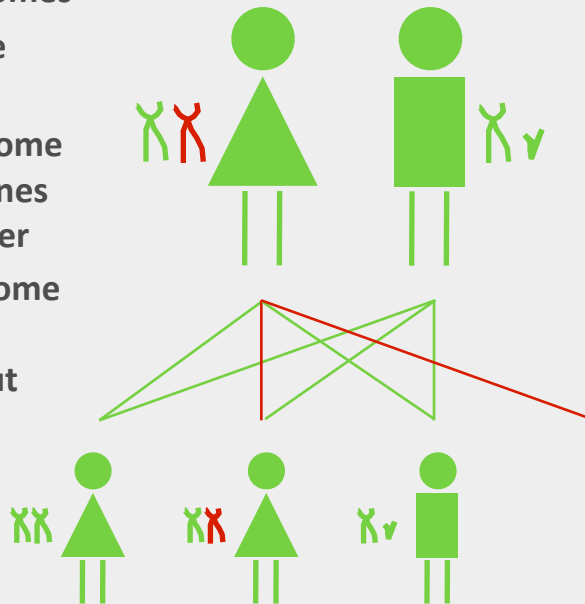
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man: one Y-chromosome  
and one X-chromosome

on the (little) Y-chromosome  
there are almost only genes  
that determine the gender

on the larger X-chromosome  
there are genes that  
determine the gender but  
also some other genes

let's say that on one of  
the X-chromosomes of  
a woman there is a  
mutated gene  
than we have four  
possibilities



woman healthy X + man X-chromosome  
= healthy girl, no mutated genes

woman sick X + man X-chromosome  
= healthy girl, one mutated gene  
the healthy X makes the girl not having  
the disease

woman healthy X + man Y-chromosome  
= healthy boy

woman sick X + man Y-chromosome  
= boy with the disease

# Heredity

---

## x-linked: on the female sex chromosome, only boys at risk

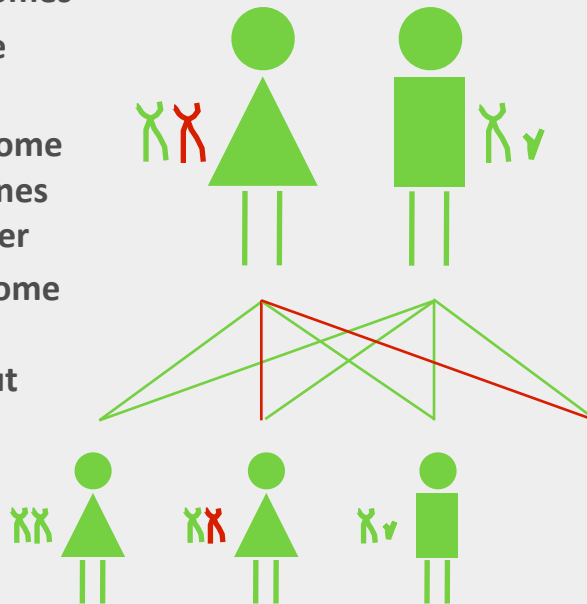
woman: two X-chromosomes

man: one Y-chromosome  
and one X-chromosome

on the (little) Y-chromosome  
there are almost only genes  
that determine the gender

on the larger X-chromosome  
there are genes that  
determine the gender but  
also some other genes

let's say that on one of  
the X-chromosomes of  
a woman there is a  
mutated gene  
than we have four  
possibilities



woman healthy X + man X-chromosome  
= healthy girl, no mutated genes

woman sick X + man X-chromosome  
= healthy girl, one mutated gene  
the healthy X makes the girl not having  
the disease

woman healthy X + man Y-chromosome  
= healthy boy

woman sick X + man Y-chromosome  
= boy with the disease

# Heredity

## x-linked: on the female sex chromosome, only boys at risk

woman: two X-chromosomes

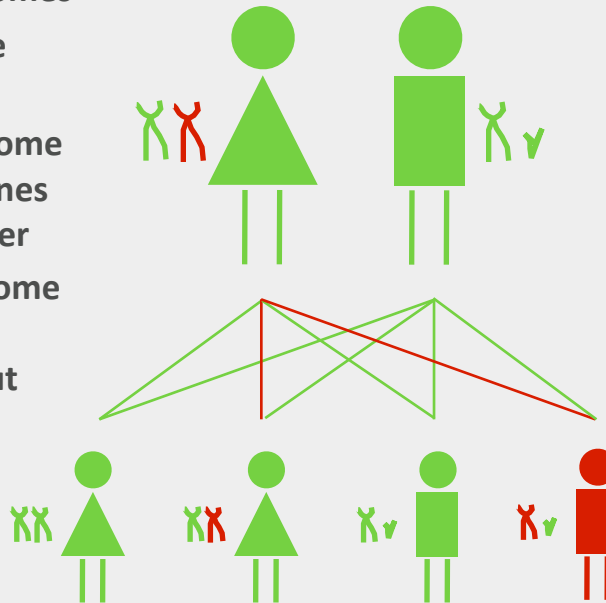
man: one Y-chromosome  
and one X-chromosome

on the (little) Y-chromosome  
there are almost only genes  
that determine the gender

on the larger X-chromosome  
there are genes that  
determine the gender but  
also some other genes

let's say that on one of  
the X-chromosomes of  
a woman there is a  
mutated gene

than we have four  
possibilities



woman healthy X + man X-chromosome  
= healthy girl, no mutated genes

woman sick X + man X-chromosome  
= healthy girl, one mutated gene  
the healthy X makes the girl not having  
the disease

woman healthy X + man Y-chromosome  
= healthy boy

woman sick X + man Y-chromosome  
= boy with the disease

# Heredity

## x-linked: on the female sex chromosome, only boys at risk

woman: two X-chromosomes

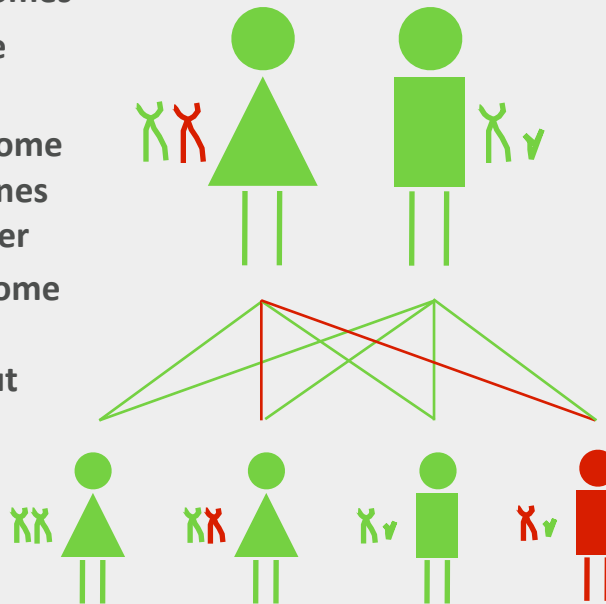
man: one Y-chromosome  
and one X-chromosome

on the (little) Y-chromosome  
there are almost only genes  
that determine the gender

on the larger X-chromosome  
there are genes that  
determine the gender but  
also some other genes

let's say that on one of  
the X-chromosomes of  
a woman there is a  
mutated gene

than we have four  
possibilities



woman healthy X + man X-chromosome  
= healthy girl, no mutated genes

woman sick X + man X-chromosome  
= healthy girl, one mutated gene  
the healthy X makes the girl not having  
the disease

woman healthy X + man Y-chromosome  
= healthy boy

woman sick X + man Y-chromosome  
= boy with the disease

girls are never affected themselves but  
half of them carry the mutated gene  
that can be passed on to her children

# Heredity

## x-linked: on the female sex chromosome, only boys at risk

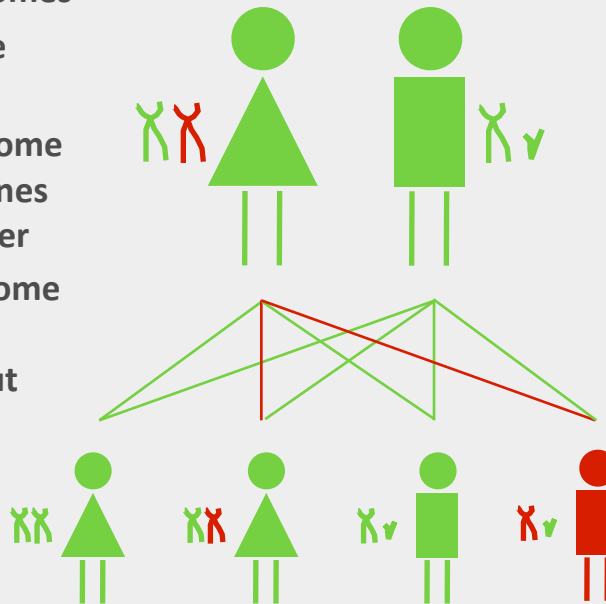
woman: two X-chromosomes

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on the (little) Y-chromosome  
there are almost only genes  
that determine the gender

on the larger X-chromosome  
there are genes that  
determine the gender but  
also some other genes

let's say that on one of  
the X-chromosomes of  
a woman there is a  
mutated gene  
than we have four  
possibilities



woman healthy X + man X-chromosome  
= healthy girl, no mutated genes

woman sick X + man X-chromosome  
= healthy girl, one mutated gene  
the healthy X makes the girl not having  
the disease

woman healthy X + man Y-chromosome  
= healthy boy

woman sick X + man Y-chromosome  
= boy with the disease

girls are never affected themselves but  
half of them carry the mutated gene  
that can be passed on to her children

boys have a 50% chance of inheriting the  
disease

# Heredity

## x-linked: on the female sex chromosome, only boys at risk

woman: two X-chromosomes

man: one Y-chromosome  
and one X-chromosome

on the (little) Y-chromosome  
there are almost only genes  
that determine the gender

on the larger X-chromosome  
there are genes that  
determine the gender but  
also some other genes

let's say that on one of  
the X-chromosomes of  
a woman there is a  
mutated gene

than we have four  
possibilities



woman healthy X + man X-chromosome  
= healthy girl, no mutated genes

woman sick X + man X-chromosome  
= healthy girl, one mutated gene  
the healthy X makes the girl not having  
the disease

woman healthy X + man Y-chromosome  
= healthy boy

woman sick X + man Y-chromosome  
= boy with the disease

girls are never affected themselves but  
half of them carry the mutated gene  
that can be passed on to her children

boys have a 50% chance of inheriting the  
disease

an example is hemophilia that ran in the  
Russian tsar family