

Guess Genes

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The Doc said a 47xxx isnâ t a type of Cowboy Hat (Darn!)

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In September of 1991, I got pregnant with my second child. I already knew my own Dadâs family tree had Spinal bifida in it. My first child tested clear of that. She's healthy, but I couldn't shake my fears. And though the babyâs daddy, Whiskeyâs family wouldnât admit to any trouble on their side â after knowing frisky Whiskey â I knew theyâ had to beâ hiding some things, besides all of Whiskey's skeletons. So I lied and told the OBGYN, Iâ d found a Downs Syndrome child closer to my family tree. Plus being 33 helped me to get the extended genetic testing. I got the test results by phone about 3 weeks later.

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Itâ s a shame weâ ve never met â knowing me and seeing me would help you see the humor in this story.

Iâ m on the phone â so the geneticist canâ t see me.â Normally you have to go in for a visit to get the results, but lady luck was on my side (or so I believed) and so I got the geneticist to give me the basic results over the phone. The baby appears to be clear, but Iâ m not. I have an extra gene, an extra â Xâ he says.

Iâ m puzzled. My mind is racing and Iâ m thinking â I know you that if youâ re missing a gene â you have Downs Syndrome. So what does having an extra gene get you? So naturally I ask, â What?â He says the test results showed that you have an extra gene. He quickly adds that girls have relatively minor negative effects with this condition. Theyâ re classified as Super Females. Thatâ s nice to know â I think -maybe I could be Wonder Woman.

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I ask him to tell me some more about this condition. You can tell from his voice heâ s delighted, but at the same time cautious when answering. Itâ s good to know heâ s experienced at this. So, he continues to tell me that scientists discovered this gene in 1959 and theyâ ve been compiling a list of the common symptoms ever since.

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*He tells me, the gene is a translocated gene, meaning both parents have to have the gene in order for their offspring to inherit it. [Iâ d learn later on â that now the scientists believe older Moms carry this gene and itâ s a sex-linked gene abnormality] When I ask about the symptoms, he adds that I may not have all of them. Some women who have this condition tend to be **mosaics**. I verbally repeat the word â **mosaics**.â My mind thinks, â Art made out of broken tiles and plaster,â but the guy quickly interjects that â **mosaic**â in the medical world means that it is likely that the woman has all the genetic material to display all the common symptoms, but for some unknown factor â she doesnâ t. â I think, Wow â Iâ m a medical Art project! And smile proudly.*

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*Â Â Â Â Â Â Â Â Â Â The geneticist starts by letting me know that Iâ m **only** a **47xxx**. But that people who have this syndrome can have even more xâ s, however I only have 3. I sigh, â Darn! I could have*

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been at least a 4x or a 5x Beaver! The Geneticist sternly informs me that a 47xxx isn't a type of Cowboy Hat. *I laugh a reassuring laugh, and add, if it was, it'd be a great Cowboy Hat.*

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Please go on. First, women who have this gene tend to grow tall early in life. He barely has time to finish his sentence, before I start chuckling. I laughed and told him *I'm 6'2" barefoot and that I was 5'4" when I was 9 yrs old, taller than my 4th grade teacher, but skinnier.* ^ *(Check! 1 for 1)*

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Please go on. Second, women who have this gene grew tall *^* but had a smaller bone structure than most women who are the same height. Again, I chuckle and tell him that my ring size was a 5 and my shoe size was a 9. *Most women my size, wear a size 8 ring and size 11 shoes. As far as tall women are concerned I am very small boned..* (Check! 2 for 2) *Please go on.*

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Third, women who have this syndrome tend to be small breasted. *Hey now!* ^ *I gasped* ^ *I know you can't see me, but I want you to know that my pregnancy has done a lot to improve that condition. I lost my position as the President of the Itty-bitty-Titty Committee due to my first pregnancy. I'm now Vice President..* ^ ^ (Check!)

I'm batting 3 for 3, what's up Doc? I thought I was a Mosaic? The Geneticist quickly added that there were more symptoms. *There's hope for me yet.*

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Fourth, women with this gene tend to have a high IQ, but are also learning disabled in the areas of Reading and Writing. (Check! 4 of 4) *I disclosed that my Mom would be pleased to hear that my learning problems weren't solely her fault, she'd always blamed herself. For years, nearly everyone believed my Learning Disability was due to the oxygen deprivation I suffered, because I drowned when I was 18 months old. Instead it was the genes. I bet that's the last time, Mom buys me Guess genes.*

That time, even the Geneticist laughed and commented that I was taking this news very well. I thanked him and asked him to continue.

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Fifth, women with this syndrome had fertility problems and lots of female problems. *It doesn't look like I'm a mosaic Doc. It took me more than 10 years to get pregnant with my first child. Besides, I've discovered I've even developed an allergy to some men's sperm.* ^ (Check! 5 of 5).

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Anymore I ask? Just one more, he said. **Sixth**, lastly, women who have this gene tend to be more emotional than most women. They cry more and they suffer from depression more. (Check! 6 of 6) *I display 100% of the commonly known symptoms for women who have an extra X chromosome. I'm ADHD (I'm not hyperactive, I tend to be a Zoner from Arizona) and I'm clinically depressed!*

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Okay if being a woman and having an extra X isn't so bad, what about men who have an extra X?
The extra X in boys is called Klinefelter's syndrome. Males who have Klinefelter's syndrome tend to be mentally retarded, have breasts, and small genitalia!

You said, my baby looked okay, right? He said, it would be best if I had an amnio-syntethsis (sp) to make sure my baby was a girl, just in case. That just in case scenario is due to all the potential Translocated-genes the scientists are still discovering!

I had that amnio. My daughter still has the scar. Later on, I had both girls genetically tested again. They both tested negative for the extra X, but the geneticists said both girls would be carriers. Both girls will need to have their own babies tested, too.

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So what message do you walk away with!

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1. Cowboy's nicknamed Whiskey may have skeletons in their closets, but seem to be genetically sound.
2. Be careful when you buy Guess genes - you never know what's in them pockets!
3. If I had known I had this gene I probably wouldn't have had kids at all.
4. Everyone carries between 6 and 20 fatal genes. **Consider it a Blessing if you and your baby are healthy.**

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Lady Jewells (2010)

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