

Doctor: Well Hello there Mrs. Andujo how are you today?

Mo: I'm....fine thanks for asking and you Doctor?

Doctor: I'm great thank you. So I just got your test results and you are pregnant with quadruplets! CONGRATS

Mo: (*panics*) what?!?!?!?!?!?

Doctor: I am kidding, (*cheesy laugh*) it lightens the mood in these situations. Or at least...that's what I was told in training... guess...not.... Anyways. I see we are here i of this special little one.

Mo: Yes Doctor (*sighs*) I am rather worried, she is only six months old but she doesn't seem like my other niece who is her exact age. There is literally only a week difference between the two girls.

Doctor: Well may I ask you for her symptoms? As in what are some things you find to be different?

Mo: Well, she isn't crawling yet like my niece and she hardly ever cries. And when she does she still smiles in between. It is confusing to me.

Doctor: Hmm well did you ever take a UBE3A screening? its the one that will tell you if there is a maternal mutation in the family genetics.

Mo: well...(pause) no doctor I never did. My husband and I are in good shape and so are our parents, so I didn't see the need to test to that extent?

Doctor: Well...from her general characteristics (the big eyes, wide smile and personality trait), I think it is angelman syndrome.

Mo: Is it a sickness or something?

Doctor: It's a neurological disorder. Almost like down syndrome but more complex.

Mo: How did it happen?

Doctor: The thing is this protein called ubiquitin protein ligase E3A gene is the one that causes it. The result of this gene being damaged or lost in the 15th chromosome is angelman syndrome. Aside from this, the disorder can also occur if two copies of the genes from the parents are given paternally, rather than having one paternal and one maternal copy of the gene. In other words if its stuck in your genetics.... there is no way of prevention its only inherited.

Mo: So this syndrome, is it common? What will happen to my daughter?

Doctor: Luckily it is only 1 in 25,000 babies who get this disease and as for your daughter, there will unfortunately be no intellectual progress from where she is now. She will be even lucky to walk and even more so if she can do simplistic communication.

Mo: Well.... not exactly the best news I want to hear... will it affect her health?

Doctor: No her health and immunity is in perfect condition, this only affects the brain and is more foreseen by actions and characteristics. At least you can tell her cheesy jokes and she will find every bit of them funny! This syndrome causes them to always be happy

Mo: Oh yes doctor (*in sarcastic tone*) something to be so thrilled about. How about her life expectancy?

Doctor: It will be completely normal. I may warn you though, at about 2-3 years she will develop seizures from the syndrome. its common but not often. I am very sorry for such news Ma'm.

Mo: Thank you so much Doctor. But as long as she's alive and well I couldn't ask for a better daughter.

Podcast Question:

1)Knowing that this syndrome is incurable and inherited what would you do if you found it to be in your genetics and you wanted to have a child with your spouse? What would your plan be?

2) If you were in Mrs. Andujo's position, what would you do to care for your child in the best way possible and be prepared for the road ahead of you? (Medical help, mental help, health care options etc)