

**Dr. David Sandberg: CAH Educational DVD**  
**Genetic Counseling Script**  
**Updated 3/27/2007 1:29 PM**

<b>Scene</b>	<b>Audio</b>	<b>Video</b>
4.2	<b>Maria:</b> Hi, I'm Maria and this is my husband, Ramon.	Maria looking at counselor
4.3	<b>Counselor:</b> Good morning. I'm Debbie Sullivan I know we spoke on the phone earlier and you had mentioned you just had a baby girl, she has CAH, and Dr. Davis thought it might be a good idea for us to meet and talk about it.	Counselor looking at couple
4.4	<b>Maria:</b> Yeah, that's pretty much how we got here. Because of Dr. Davis. He said you'd be able to explain some things about CAH.	Maria looking at counselor
4.5	<b>Ramon:</b> This is all really new to us. And I know Maria made the appointment with you, but what, exactly is it that you do?	MCU of Ramon
4.6	<b>Counselor:</b> Okay, I'm a genetic counselor. Genetic counselors have a background in biology, genetics and counseling. Genetic counselors typically come from a Masters degree genetic counseling program, but other qualified and certified health care professionals such as Masters degree genetic nurses or physicians also provide genetic risk assessment and genetic counseling.  What we do is meet with individuals and families who are affected by a genetic disorder like CAH, or when there is a genetic condition	MCU of counselor  (cut to reaction shots)

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	<p>suspected in the person or family. We assess what the condition is, and discuss its cause, the associated features, and the medical problems or concerns.</p> <p>We explain and help people understand the technical &amp; genetic aspects of inherited disorders - like the genetic reason a person has a certain disorder and possible genetic testing. We help advise and connect the person with professionals and resources they need to deal with who treat or manage the condition. We discuss how this may impact other relatives or future generations, and assist families in coping with this information.</p>	
4.7	<p><b>Ramon:</b>            Oh, so you know things about how our daughter got this? And if we have another baby, if they'll have it too?</p>	MCU of Ramon
4.8	<p><b>Counselor:</b>            Yes and no. CAH is inherited, so it's a combination of genes that caused your daughter to have CAH - and we'll talk about that in just a minute. And we can talk about the likelihood that your next child could have CAH. But no one can say, for certain, if they will or won't.</p>	MCU of counselor
4.9	<p><b>Maria:</b>            Okay, then how did she get it? I did all the prenatal things right - the appointments and vitamins. I watched all the pregnancy videos. I can't figure out what I did wrong. But my family, on my father's side has a lot of bad genes. My uncle has diabetes and my dad's mom had it. And her husband died of a stroke or a heart attack or something when he was young.</p>	MCU of Maria
4.10	<p><b>Ramon:</b>            My side of the family has been trying to figure out where it came from. Unfortunately, and I think it's really getting to Maria, they're blaming everybody else for it. They figure that since no one has it on our side, it</p>	MCU of Ramon

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	must be from her side. It's been horrible. I want them to be a part of little Tiffany's life, but it's such a mess. How did this happen? How do I explain this to them?	
4.11	<b>Counselor:</b> For CAH, we're talking about genes that are specific to CAH. So you can feel sure that there's nothing either of you did that "caused" her to have this condition. A baby doesn't have CAH because a parent did or didn't do something during pregnancy. Also, it's not caused by how you take care of your baby once he or she is born. It's all in the genes.	MCU of counselor
4.12	<b>Ramon:</b> It's been awhile since my high school bio class... And we've heard so many words that they almost don't make any sense anymore...	MCU of Ramon
4.13	<b>Maria:</b> Nothing makes sense anymore. We were supposed to have this happy, healthy little girl. It was the worst day of my life, when it should have been the best (crying), but the doctors tell me she's doing really well. I have 2 boys who are fine – they don't have it. They don't show any symptoms and they're growing fine and they just love her and she's doing well.	MCU of Maria
4.14	<b>Counselor:</b> It's a lot to take in all it once. Sounds like you felt pretty overwhelmed.	MCU of counselor
4.15	<b>Ramon:</b> Yeah, we've been doing our best to hang on. To try and figure out what's going on. To take it all one day at a time.	MCU of Ramon
4.16	<b>Maria:</b> It's been a couple of months now, but we seem to keep going between feeling like we can handle it all and that we know everything that we need to know, then something will happen. Something tiny like... I don't know... But like just last week I was all upset again and it didn't	MCU of Maria

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	<p>make any sense. I was feeling all angry. And I know that's not right. I shouldn't be angry. Especially not at her. It's not like it's her fault.</p> <p>It's weird though: it kind of comes in waves. Sometimes I feeling really worried or anxious and then it goes away. Well, not really goes away, just takes a back seat and then I can understand what's going on and make some good decisions and then I get upset again...</p>	(cut to reaction shots)
4.17	<p><b>Ramon:</b>          She can't even think straight. But it's not like it's just her. I get that way too.</p>	MCU of Ramon
4.18	<p><b>Counselor:</b>          A lot of people feel angry, shocked, scared, or overwhelmed. It's okay to feel that way. It's normal to feel okay one moment and then angry the next. And it can be difficult understanding all the new information and even more difficult trying to use that information to make decisions, especially when you're feeling upset. What matters for you and your family is what you do to cope with it. When you first learn that your child has CAH, you don't just all-of-a-sudden understand everything about it and figure out how to cope with it; instead it's a process. And it takes time. It's okay to allow yourself some time to deal with it all. What do you do when you're feeling upset?</p>	<p>MCU of counselor</p> <p>(cut to reaction shots)</p>
4.19	<p><b>Maria:</b>          Sometimes I listen to my favorite music. I sit with her there in my arms and just listen. It really helps me clear my head. Just holding her. Ramon goes and tries to find information.</p>	MCU of Maria
4.20	<p><b>Ramon:</b>          I get on the Internet a lot. I make lists of things I need to do, things I want to know, questions for the doctors. I feel like I'm more in control.</p>	MCU of Ramon
4.21	<p><b>Maria:</b>          I called my mom and asked her to come with me to Tiffany's doctor's</p>	MCU of Maria

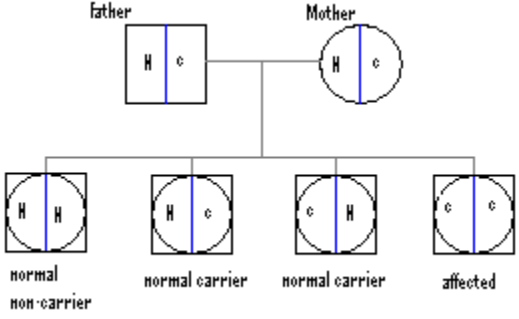
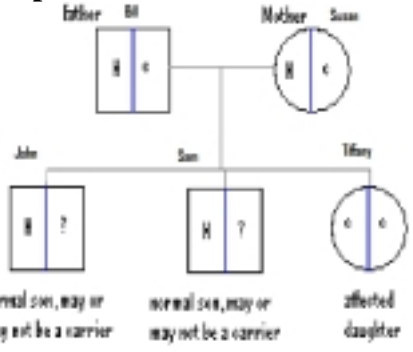
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	<p>appointments - especially when Ramon can't be there because of work. She helps take care of her a lot and sometimes she can pick up on things that I didn't hear the doctor say the first time around.</p>	
4.22	<p><b>Counselor:</b>          Sounds like you're on the right track. There's a word we use to describe what you're doing: using <b>COPING STRATEGIES</b>. Coping strategies can be broken down into two groups: problem-focused and emotion-focused. <b>PROBLEM-FOCUSED</b> strategies are best used to change circumstances; for example, calling your mom to help or making an appointment for your daughter to see the doctor. <b>EMOTION-FOCUSED</b> strategies are used to decrease emotional distress; for example, listening to music, taking a walk, or talking to a close friend or family member. And there are some things, like finding out information, that fall in both categories because they address both aspects</p>	<p>MS of counselor           (superimpose graphics of highlighted terms)</p>
4.23	<p><b>Ramon:</b>          How about we do some of that "finding out information" thing while we're here? You were saying something about how CAH is genetic...</p>	<p>MCU of Ramon</p>
4.24	<p><b>Counselor:</b>          Right, let's start with genes. Genes are the basic building blocks that contain information about an individual's characteristics like eye color and height, but also information important to functions of cells. Genes tell cells to make different proteins. If there is a problem (a mutation) in a gene, or pair of genes, it can cause or increase the likelihood for a health problem like heart disease, or CAH.</p> <p>Genes are located on chromosomes within the nucleus (central structure) of cells -- we have a total of 46. These are in 23 pairs -- 22 pairs of autosomes that are the same in men and women, plus 1 pair of sex chromosomes -- 2 "X's" in women and 1 "X" and 1 "Y" in men. When a person is conceived, they inherit one copy of each chromosome</p>	<p>MCU of counselor   <b>Illustrations/graphics:</b>          Chromosomes</p>

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	<p>pair – and the genes on those chromosomes -- from the mother’s egg cell and similarly one copy of each from the father’s sperm cell. So Tiffany inherited half her genes from you, Maria, and the other half from you, Ramon.</p> <p>For each characteristic, a child gets one gene from the mother and one gene from the father. With CAH, typically both parents of the child with CAH are carriers for an alteration in the gene for that condition. The normal CAH gene makes a protein important to cell function, the 21-hydroxylase enzyme (remember from the Medical Management chapter that it is the 21-hydroxylase enzyme that affects production of cortisol, aldosterone, and androgens).</p> <p>We also know that to have CAH the person must have 2 copies of the problem gene for CAH (the one that affects the 21-hydroxylase enzyme) because the condition is recessive. <i>Recessive</i> means that your child needs to have two copies of the affected gene in order to have CAH. This would be the case for your family. Neither you, Maria, nor you, Ramon, have CAH, yet your daughter does. That means you must both be carriers, or what is called <i>heterozygous</i> for this trait.</p>	<p>Recessive: requires both copies of the gene to show that feature</p>
4.25	<p><b>Maria:</b>            So, we're both carriers of CAH. And then that means that on both sides of our family, there have to be others who are carriers...otherwise, how else would we be?</p>	MCU of Maria
4.26	<p><b>Counselor:</b>            Precisely. One way of looking at it is like this. Here, we use boxes for males and circles for females. The father and mother are on the top row, and children are on the bottom row. Since we're just talking about probabilities now, the children are represented by both a box and a</p>	<p>MCU of counselor</p> <p><b>Graphics:</b></p>

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	<p>circle, since they could be either boys or girls. We also use "N" to mean the “normal” (or non-affected) gene and "c" for recessive (the affected CAH gene). Here, each parent is “N” ”c”</p> <p>As you can see, when both parents are carriers for the gene alteration, the chance of having a child with CAH is 1 in 4, or 25%. That means that there is a 3 in 4, or 75% chance that your child would <b>not</b> have CAH. You can also see that, there is a 2 in 4, or 50% chance that you would have a child who, like you, is a carrier for CAH.</p>	<p style="text-align: center;"><b>Video</b></p>  <p style="text-align: center;">father: H c      Mother: H c</p> <p style="text-align: center;">     normal non-carrier    normal carrier    normal carrier    affected   </p>
4.27	<p><b>Maria:</b>        Okay, so that box [pointing] would be Ramon, and that circle would be me. And then our kids are...wait, we only have three kids.</p>	MCU of Maria
4.28	<p><b>Counselor:</b>        This picture just shows the possibilities for you and Ramon. That you have a 1 in 4 chance of having a non-affected, non-carrier, 2 in 4 chance of having a non-affected carrier, and 1 in 4 chance of having an affected child.</p> <p>If we made the same type of drawing specifically for your family, the picture would look like this one.</p>	<p><b>Graphic:</b></p>  <p style="text-align: center;">father: Ramon    Mother: Maria</p> <p style="text-align: center;">     John                      Sam                      Tiffany      normal son, may or may not be a carrier    normal son, may or may not be a carrier    affected daughter   </p>
4.29	<p><b>Maria:</b>        Oh, okay. So that’s Ramon, that's me, that’s John, that's Sam, and</p>	<b>Graphic, continued</b>

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	Tiffany is that little "cc" in the circle...	
4.30	<b>Counselor:</b> Right	
4.31	<b>Ramon:</b> And our sons might be Nc or NN, that's why you have the "N" and the question mark - because they might be carriers or they might not be affected at all - like not even be a carrier?	<b>Graphic, continued</b>
4.32	<b>Counselor:</b> Yes, they may not even be carriers. However, even if one or both are carriers, this does not pose a health problem for them – just like with both of you.	MCU of counselor
4.33	<b>Ramon:</b> Well, what about if we had another child? That would be a total of four children and if only one in four gets it, then the next one will be safe, right?	MCU of Ramon
4.34	<b>Counselor:</b> It doesn't quite work out that way. A lot of people wish it would, but it doesn't. What the 1 in 4 chance of having CAH means is that with <u>each</u> pregnancy the two of you have, there is a 1 in 4 chance. So John had a 1 in 4 chance of having CAH and a 3 in 4 chance of not having it - it turns out that he fell in the 3 in 4 group of not having it. Same thing with Sam. And the same thing for Tiffany, except she just happened to fall in the 1 in 4 group. We can't tell if your next child would fall in the 1 in 4 group or the 3 in 4 group.	MS of counselor
4.35	<b>Ramon:</b> So it's kind of like how there's a 50-50 chance of either a boy or girl, and just because we have two boys and one girl, it doesn't guarantee	MCU of Ramon

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	that we'd have another girl.	
4.36	<b>Counselor:</b> Exactly.	MCU of counselor
4.37	<b>Maria:</b> I heard there was some sort of medication they give pregnant mothers that cures CAH - so that could work too, right?	MCU of Maria
4.38	<b>Counselor:</b> It's not quite like that. The medication you heard about is called <i>Dexamethasone</i> [DES6]that some mothers choose to take during pregnancy. If the mother begins taking it at the right time during her pregnancy, it reduces the extent of changes to the external genitalia of affected girls. But it doesn't cure CAH. All it does is keep a girl's genitals from looking different from those of other girls.  I also want you to know that using this medication is not without controversy. If you are considering or do become pregnant, I highly recommend consulting with medical doctors about using any medication like Dexamethasone, especially since you'd need to take it before it is possible to do the tests to find out if the baby is a boy or a girl or if the baby had CAH. It is also important to talk to your doctors and/or a specialist about the various medical aspects of CAH, how to manage these, and how features of CAH may vary from family to family	MS of counselor  Superimpose graphic: Dexamethasone
4.39	<b>Maria:</b> Oh, so we're back to thinking about 25% chance of having it and 75% chance of not having it. But how about Tiffany? What if she had children?	MCU of Maria
4.40	<b>Ramon:</b> Wait. Can she have children? You know, because of all the medications and how she looks down there?	MCU of Ramon

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4.41	<p><b>Counselor:</b>            I don't know the specifics of your daughter's condition, but I can tell you about what we know about most girls...well, I guess at that point I should say "women"...with CAH. Most women with CAH can become pregnant. Just like other women, they can carry their babies to term; but, usually they'll need to deliver by c-section.</p>	MCU of counselor
4.42	<p><b>Ramon:</b>            Oh, that's right, Dr. Davis did say something about that. It's just hard to put it all together.</p>	MCU of Ramon
4.43	<p><b>Counselor:</b>            That's okay. You're right; it is a lot of information. And it's okay to go back over it as many times as you need to and to keep asking questions until it all makes sense.</p>	MCU of counselor
4.44	<p><b>Ramon:</b>            Thanks. So Tiffany... will she have kids with CAH?</p>	MCU of Ramon
4.45	<p><b>Counselor:</b>            Actually, it is most likely her children would not have CAH. First, remember that you need two recessive genes for CAH - one from each parent - in order to have a child with CAH. Her children would definitely inherit one recessive CAH gene from her...</p>	MCU of counselor
4.46	<p><b>Ramon:</b>            That's because she only has recessive genes to give.</p>	MCU of Ramon
4.47	<p><b>Counselor:</b>            Yes. And the only way she could have a child with CAH is if she partnered with another person who either has CAH or is a carrier. Statistically speaking, it's unlikely for a person with CAH to have a child with another person who either has CAH or who is a carrier. Based on estimates from the general population, about one person in every 50 or 60 is a carrier and has one CAH gene mutation. When the</p>	MCU of counselor

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	geneticists do the math, they find that the chance of your child having a child with CAH is about 1 in 100.	
4.48	<b>Ramon:</b> Can someone get tested to see if they are carriers? Like someone she's thinking about for a husband?	MCU of Ramon
4.49	<b>Maria:</b> Or our sons? Can they get tested?	MCU of Maria
4.50	<b>Counselor:</b> Yes they can. There are two possibilities. One is a test, called a "Synacthen [DES7]stimulation test," that is used to figure this out. But doctors debate about how reliable it is. The other way, that is more reliable, is genetic testing. It involves a molecular genetic test of the gene associated with CAH (it's called the CYP21A2 gene) for problems like common changes and alterations. If this is something you would like to do, let's talk about it some more.	MCU of counselor
4.51	<b>Maria:</b> Okay... it's probably not even going to be relevant until they're all grown up anyway...	MCU of Maria
4.52	<b>Counselor:</b> Right. You already know your sons don't have CAH, so learning if they carry the gene isn't going to change how you or their pediatrician care for them. So it might just be a good idea to remember that there is a test, and when the time is right, you can do it.	MCU of counselor
4.53	<b>Ramon:</b> Yeah, that makes sense. I'm glad we had the chance to talk about everything and make sense of it.	MCU of Ramon
4.54	<b>Maria:</b> You've definitely cleared up a lot of things for us. I'm actually feeling a bit better than when I walked in here today.	MCU of Maria

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4.55	<p><b>Ramon:</b>            Could we take a copy of that diagram with us? I think it would help to look at it...to recall what we discussed today and to explain to our families. Can they call for information or genetic counseling?</p>	<p>MCU of Ramon</p> <p>Link to “Instructions for Accessing Documents”</p>
4.56	<p><b>Counselor:</b>            Sure. Are they all local?</p>	<p>MCU of counselor</p>
4.57	<p><b>Ramon:</b>            About half and half. Half live out of the state.</p>	<p>MCU of Ramon</p>
4.58	<p><b>Counselor:</b>            All right, let me jot this down for you [writing]. It's the National Society of Genetic Counselors (NSGC). If you, or your family, go to their website (<a href="http://www.nsgc.org">http://www.nsgc.org</a>), you can find a list of genetic counselors in their area.</p>	<p>MCU of counselor</p>
4.59	<p><b>Maria:</b>            Thanks. I'll probably pass this on to my brother...well, everyone, but especially him because he's thinking about starting a family and I figure if I'm a carrier, he could be too and not know.</p>	<p>MCU of Maria</p>
4.60	<p><b>Ramon:</b>            Yeah, thanks. When we get home, if we have a question, would it be okay to call?</p>	<p>MCU of Ramon</p>
4.61	<p><b>Counselor:</b>            Sure. As we were saying earlier, it's a lot of information and it takes time to make sense of it all and feel comfortable with it. And it's important that you do feel comfortable with understanding what's going on for you and your family - so please do call when you need to.</p>	<p>MCU of counselor</p>

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