

About Health TV with Jeanne Blake
MSA
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JEANNE BLAKE: I'm Jeanne Blake. Imagine being in the prime of your life and suddenly you notice a change in the way that you feel. You're not as steady as you once were. You feel dizzy or faint when you stand up, or your speech may be slurred. These can be symptoms of a disease that until recently has gotten little attention. It's called Multiple System Atrophy, or MSA. On this edition of *About Health TV* we'll learn about MSA with the help of a neurologist from Boston's Beth Israel Deaconess Hospital, Dr. Roy Freeman. He is also an Associate Professor at Harvard Medical School. Dr. Freeman, thanks for joining us and I should mention that a little later we'll be joined by Rob Langer and his wife Carol Robb, is living with MSA. Tell us about this disease. What is it, multiple system atrophy?

DR. FREEMAN: Well, muscular system atrophy, as the name implies, is a disease that involves multiple systems within the nervous system. The most important systems that are involved are those that involve movement, balance, coordination and even more importantly the autonomic nervous system, which is the part of the nervous system that works automatically.

JEANNE BLAKE: Okay, so what you're saying – we'll go into each one of those so if someone is listening and they might identify with some of the symptoms – but it's a disease of the brain. When someone is living with MSA, what's going on inside their brain that's causing the trouble?

DR. FREEMAN: Those systems, the motor systems and the autonomic system is slowly degenerating. The nerve cells that are involved in those functions are slowly dying.

JEANNE BLAKE: Now you're talking about autonomic. What is our autonomic system?

DR. FREEMAN: Our autonomic nervous system is the part of the nervous system that works automatically. It works without you thinking about it. It controls blood pressure, heart rate, bowel, bladder and sexual function.

JEANNE BLAKE: Okay, but we don't know today still what causes this.

DR. FREEMAN: We have no idea. We are beginning to get closer to understanding parts of the process which causes these cells to die, but we still do not have a definitive answer.

JEANNE BLAKE: And what is that? What is part of what causes the cells to die?

DR. FREEMAN: Well, we have discovered that there are a number of abnormal proteins which may play an important role in the cell death.

JEANNE BLAKE: Okay, so who's at most risk for this disease?

DR. FREEMAN: Well, people in their middle age. Males and females, but males slightly more than females but in the middle ages.

JEANNE BLAKE: Let's talk about the symptoms because they really – the diagnosis of this disease is really challenging and people present a wide range of symptoms that can be a lot of other things. You broke it down into four parts. Let's revisit those. You talked about that there are symptoms that deal with movement. Let's just go over what some of those can be please.

DR. FREEMAN: Well, those are related to balance, people will not be able to maintain their balance as well as they used to, they will move more slowly.

JEANNE BLAKE: All the time or sporadically?

DR. FREEMAN: All of the time.

JEANNE BLAKE: All of the time, okay. I mean, suddenly you just wake up one day and you're moving more slowly?

DR. FREEMAN: Coming on very gradually and interceptively so balance, coordination, our fine movements, even control of the voice will deteriorate.

JEANNE BLAKE: Over what period of time?

DR. FREEMAN: Well it will occur very, very gradually over a period of years.

JEANNE BLAKE: Give me an example of how confusing this can be to a patient. We'll hear from Rob and Carol about this a little bit later, but I'm wondering if you can give me an example of a patient that you've seen that came in and said, "Gee, I don't know what this is."

DR. FREEMAN: Patients will report that they used to be very agile, were able to participate in sporting activities, carry out fine kind of work, writing, and then they will note that over a period of time these abilities will slowly become worse. They will have a tendency to fall, they will feel off balance, abilities to move, even drive or dance or carry out sporting activities will decline.

JEANNE BLAKE: Okay. Now one of the really big red flags is the sudden loss of blood pressure. So I made a little note to myself to ask you about that and I referred to it in leading into the program about a sudden dizziness or even feeling faint or fainting when you stand.

DR. FREEMAN: That's right. Blood pressure control is one of those automatic functions. When we move from lying down to standing up, the nervous system needs to make a series of very rapid adjustments in order to increase blood pressure or maintain blood pressure against gravity. And with this disease, that ability to maintain blood pressure when upright gradually declines to the point of, even in some patients being unable to move from a lying down to a standing up position, and many patients are forced to spend time in bed, prolonged periods of time in bed because they're unable to stand without fainting.

JEANNE BLAKE: I see. It's starting to make some sense to me because that part of the brain that makes all of that automatic is losing brain cells, therefore, losing its ability to do all that.

DR. FREEMAN: That's quite correct.

JEANNE BLAKE: All right. Let's move on to the bladder and the bowel problems, which is another indicator.

DR. FREEMAN: In exactly the same way that part of the brain in the nervous system which controls those automatic bladder functions and bowel functions also decline and deteriorate. So individuals with this disease are unable to move their bowels, unable to move their bowels, unable to empty their bladder in the automatic way that we do those processes.

JEANNE BLAKE: Okay. And the last one, which Rob will tell us about which was one of his first symptoms is the sleep disturbances.

DR. FREEMAN: This was actually until fairly recently an unrecognized aspect of this disease. There are a number of sleep problems that occur with this, including very heavy breathing, loss of breathing during sleep so that the kind of breathing that we do while we are sleeping which occurs totally automatically may even disappear when patients will have prolonged periods of breathlessness while sleeping, the vocal chords will be discoordinated so the vocal chords can automatically close during periods of sleep. Patients will have very, very vivid dreams and they will even act out their dreams during sleep. Now this was until fairly recently unrecognized, but as time has gone by more and more physicians are aware of this manifestation.

JEANNE BLAKE: What do you mean, Dr., acting out a dream? Give us an example.

DR. FREEMAN: There are people who, as part of their dream will imagine that they are moving their arms, having a fight, and rather than this just being a passive, mental process, with this disease often they will actually very aggressively move their arms, hit their spouse, fall out of bed. So this is what is known as, and I'm going to use a medical term, REM behavior disorder. And so that during the REM period, the rapid eye movement period of sleep, the dreams which usually are passive can, in fact, be quite aggressively acted out.

JEANNE BLAKE: The other symptoms that we've talked about made more sense to me in terms of being logical, but I don't get this one. Do you know – has there been research into what the REM behavior disorder is caused by within the brain?

DR. FREEMAN: Well usually during the REM period, the body is paralyzed, doesn't move. The nervous system in a way induces that paralysis. It's probably a way that we protect ourselves during that period of vivid dreaming. When those nerve cells die, the ability to paralyze the nervous system is lost so REM sleep dreaming becomes an active process.

JEANNE BLAKE: Okay, now I understand. Thanks. In terms of diagnosis, I read some of the literature about this, and it can be a nightmare for someone trying to get a diagnosis. How many of the symptoms does one have to present with before someone or a doctor will say this could be MSA?

DR. FREEMAN: I think, and this is a point that cannot be made strongly enough. Here you have a situation where the most important functions of the body, the most vital functions - blood pressure, bowel, bladder - are gradually deteriorating and it's so clearly recognized. And so many patients for many years are regarded as having psychiatric problems that when these patients present these physicians who are unaware of this entity which is relatively common, not a rare disease, they are regarded as having psychiatric problems, referred to psychologists and psychiatrists. Most patients will give horror stories as to how they have seen doctor after doctor, even neurologists, before the diagnosis is made.

JEANNE BLAKE: It's often confused with Parkinson's because a lot of the symptoms mimic Parkinson's.

DR. FREEMAN: That's absolutely correct.

JEANNE BLAKE: Do some patients think for months or years that they have Parkinson's when actually they have MSA?

DR. FREEMAN: Those in some ways are even the fortunate patients. There's patients that present Parkinson's-like problems I think have a more happy course because at least they are then given a neurological diagnosis. But you're quite correct. This is in many ways confused with Parkinson's Disease. The big difference is that the medications that we use to treat Parkinson's Disease don't work quite as well with this, and the autonomic problems – those automatic functions are much, more worse.

JEANNE BLAKE: And in fact, Aldopa, which is a common treatment for Parkinson's, doesn't work for MSA and that's one way of diagnosing a patient, right?

DR. FREEMAN: That's to some extent correct. It doesn't work quite as well. It works. The period of time for which it works is much, much less and the effect is not nearly as dramatic.

JEANNE BLAKE: How does a diagnosis occur? Someone comes to you and first of all they're lucky because they're coming to one of the medical centers that knows about this. If someone goes to – let's say that they come to you because that's how they'll get to the diagnosis rather than if they go to a doctor who doesn't know about it then they get shuffled around and we've covered that. If someone comes to you, how do you start to determine whether it's MSA, Parkinson's, or something else?

DR. FREEMAN: The combination of severe autonomic problems, be it blood pressure, bowel, bladder, sexual dysfunction, in combination with the movement disorder at the upset of the disease is characteristic of this entity and there is nothing else like that. Parkinson's Disease does not present like that. With Parkinson's Disease there are mild movement problems occurring early with very little by way of autonomic problems.

JEANNE BLAKE: On Saturday I was happy that you invited me to attend a national conference. It was educational for one thing, but it was interesting to see so many different people from all over the country who are living with MSA come together in one room and talk about some of the issues that they face and, indeed, one of them is getting the word out about this. And over the last ten years I can tell from the charts in the side shows, it has become much more widely recognized. Why do you think that is? What are the reasons for that? The science hasn't really improved, I mean, there aren't really great treatments yet, so I'm wondering what you attribute that to.

DR. FREEMAN: The science has improved to some extent. I think our understanding of the disease has improved quite substantially, not nearly enough and we need to put a lot more effort and we need to have a lot more support from a variety of sources in order to develop our understanding of this disease. I think the awareness to answer your question has come from several areas. It's come to a large extent from patient advocacy and I think one of the big changes in many diseases over the past five years has been the extent to which patients have driven the awareness of diseases such as this. I think physicians also have become more aware of it due to those of us who have in our expertise an interest in the sanctity and have publicized it, but I think there's a lot more work that needs to be done in this area.

JEANNE BLAKE: Right, and you're publishing papers and attending conferences and speaking about it.

DR. FREEMAN: Of course.

JEANNE BLAKE: So when I speak about the science not being as advanced, I think what I meant is that patients who are living with this still don't have a lot of options for treatment.

DR. FREEMAN: There are options for symptomatic treatment and I think we've progressed quite substantially in that field. I think we've learned better, not well enough, as to how to control the blood pressure problems, the bowel problems, the bladder problems and so on. I think what we are working

with now is to make those treatments more effective and also, even more importantly, to arrest the course of this disease.

JEANNE BLAKE: So one can move more comfortable with the disease, but there's still not the progress that you'd like to see towards your ability to be able to tell a patient that there is a cure or that they won't succumb to this disease within an average of 10 years.

DR. FREEMAN: That's correct.

JEANNE BLAKE: But there are patients who live for 20 years.

DR. FREEMAN: That's absolutely right, and I think we need to improve our methods of diagnosis, we need to improve our methods of treatment and, most importantly, we need to arrest the progress and prevent this disease.

JEANNE BLAKE: Well you're hard at work at it. Dr. Freeman, thanks for talking with us about MSA. We appreciate your insight and your expertise today. And we will be right back and we'll be joined by Carol and Rob Langer, who tell us about living with MSA in a moment.

JEANNE BLAKE: We continue our discussion about MSA, Multiple System Atrophy, with Rob and Carol Langer. Rob is living with MSA and was diagnosed how many years ago, Rob?

ROB: It was 1998.

JEANNE BLAKE: 1998. So you've lived with this disease for four years. What you're your first symptoms?

ROB: Well there's the symptoms I was diagnosed on, and they're the ones that existed pre-diagnosis that we now recognize were indications I had the disease.

JEANNE BLAKE: And what were the earliest symptoms?

ROB: The earliest ones were fainting, dizziness.

JEANNE BLAKE: So it really affected your balance early on.

ROB: Yes.

CAROL: I think the earliest symptom that appeared was back in the early nineties where Rob presented with what's known as a REM behavior disorder where he was acting out his dreams.

JEANNE BLAKE: That's what Dr. Freeman was talking about.

CAROL: Right, exactly, and we had no idea that there way anything more to that than we thought was sort of a wild and crazy sleep disorder. He would talk, we could have whole conversations in his sleep. He would get out of bed, I had bruises a few times where he would whack me, but he was given medication for that which calmed it down substantially and sort of, life tooled on.

JEANNE BLAKE: Rob, you had a very high power job as a Chief Financial Officer for several companies but eventually you had to give up your work.

ROB: One of the things it does, it's talked about very much is the impact on your cognitive capabilities of the disease and your body slows down as time goes by and it slows it slows your brain. I find that I'm not as able to formulate ideas or use bring order to a bunch of items on a piece of paper or something like that.

JEANNE BLAKE: There are stories of people who have sought a diagnosis for years and you were in that sense lucky that you were in Boston with a major medical institution such as the Beth Israel that does have expertise and Dr. Freeman. Rob, you're wanting to say something, go ahead.

ROB: Well I was just going to say that we're extremely fortunate from support groups around SDS patients. I think I was really very fortunate to have the doctors that I had.

JEANNE BLAKE: Rob has just referred to SDS, which we're really going to confuse you. That's Shy-Drager Syndrome, which is one of three ways that MSA can manifest. Carol, you've got a great analogy for explaining how MSA does present in three different versions.

CAROL: Well, I have to give credit for this to our support group members, but the analogy is that of a carton of Neapolitan ice cream, you have the three flavors – chocolate, vanilla and strawberry – and if you think of MSA as the whole carton, and you think of the autonomic, what's often known as SDS, as being one of those flavors, the cerebello or the balance issues as being another, and the

Parkinsonian problems or the rigidity and some of the speech things as being another. Depending on the luck of the draw, and it's hard to think of luck as being part of this, but depending on what happens to you initially you may have a predominance of one of those flavors, if you will. And in Rob's case, his symptoms are – his early symptoms anyway – are primarily autonomic. So he's got what's known as SDS, which is just a variation or a kind of multiple system atrophy. The sad thing, of course, is that at the end of the day as this progresses, you eat the whole carton. You end up with all three flavors. So ultimately he'll have all those.

JEANNE BLAKE: And that is a frightening prognosis for you to live with every day, I'm sure. There is a support group nationally and I'm fascinated by the fact that it's an online support group and many of the folks that were there are on Saturday participate in that. Have you done that, Rob? Have you participated in the national support group?

ROB: Not as much as Carol, certainly.

JEANNE BLAKE: Okay. What's that like, Carol?

CAROL: Well it's really an incredible experience. If there's one good thing that's come out of Rob's illness, it's just an incredible outpouring of support and strength from people around the world. There are almost 700 people on the internet support group, and they share the joys and sorrows of living with multiple system atrophy and they gave me a huge amount of knowledge. There's all sorts of information posted on the web on a regular basis; every research paper we can get our hands on gets posted. But it's a place where people can go and if their loved one or patient is having a problem, they can say we're having a problem with balance, has anyone found a good treatment yet?

JEANNE BLAKE: And they'll get 700 responses.

CAROL: Well it's interesting. They all must have 100 people, but probably only 50 of them are active on a regular basis but there are – the most touching postings to me are the people who will say my loved one just passed away and I've never written to this group before but I want you all to know that I've been reading your list for five years and you've been such a source of support and strength to me and my family and even though you don't know me I want to thank you all.

JEANNE BLAKE: Why is that kind of support, particularly with an elusive and degenerative disease like this, so desperately needed for both the patient and the caregiver?

CAROL: I'll take a first shot at that, but I'd say because it's so isolating. There were people at the meeting on Saturday that – there was one man who came up to me and said this is wonderful, I've never seen another person with this disease before.

JEANNE BLAKE: Wow, that's incredible.

CAROL: And you get diagnosed with this thing and it's like, what? I have what? What is it? What do you do about it? You know, that was sort of our first reaction, it's like, okay.

ROB: You go home and you start to find out.

JEANNE BLAKE: You put the pieces back together and you cope and you move on, but what do you think is the inspiration under all of that that's kept the two of you going and close and connected and wanting to continue to make a difference?

JEANNE BLAKE: Are you and Rob trying to make a difference in the ways that he's just described, Carol?

CAROL: Well, I think there are a number of ways. I am going to talk a little bit about the diagnosis because we can sit here and talk about this very rationally at this point, but it's a real punch in the gut when you get this diagnosis. When we sorted out the pieces and what this meant, we spent the first probably two weeks just crying. Every time we looked at each other we would just be in tears. And you slowly kind of go, well I suppose we can spend the next "x" years doing this, and we still do sometimes. Or we can sort of go on with it. Let's pick up where to go from here and you say, okay what do we do with this? And they say we can treat the symptoms and the symptoms with – Rob's been extremely fortunate that his symptoms are being managed by medication but the symptoms are pretty awful. But there's nothing to slow it down, there's nothing to treat the cause. Those brain cells are dying as we speak, and that's a pretty awful thing. One other thing that we've done is getting involved with the support group, the national support group, and we're trying to help get the word out that this disease exists, that there is help at least for the symptoms because so many people do go for years and the stories I've heard on the national support group are hair raising. It's like, I went from one doctor to another, they told me it was all in my head, it's sort of a joke, you know, they told me it was all in my head, ha-ha, I guess it really is. But it's physical; it's not mental. And we also tried to give back to the people who are on the internet support group. We look to them for help and we give help back to them.

JEANNE BLAKE: Well, you sure do and I want to thank you for coming in today and talking with us about it. You're an inspiration, and I know that you're touching a lot of lives and I hope that through this program you'll be able to reach some more. I said this on Saturday and I'll say it again. If we can help in any way, we will. We wish you the best and thank you for joining us for this edition of *About Health* TV. I'm Jeanne Blake. I'll see you next time.

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