

Genes and confidentiality; sore throats and cancer; diet for epilepsy; shaving for hospital drips

Genetics and confidentiality; a fascinating legal case where a woman is suing the hospital trust that looked after her father with Huntington's disease for not warning that she too could be affected.

And a well established use of very low carb diets that isn't so well known - to treat complex childhood epilepsy. Plus cancer of the voice box and persistent sore throat. And should hairy arms be shaved for a hospital drip? This question has prompted a transatlantic spat when Sir Andy Murray posted a photograph after his recent hip operation.

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INSIDE HEALTH - Programme 5.

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Porter

Hello. Coming up today: Sore throats and headlines prompted by new research suggesting that persistent soreness can be sign of cancer of the larynx.

To shave or not to shave, that is the question prompted by Andy Murray's hairy arms in a photograph taken after his recent hip operation. Margaret McCartney has been investigating the pros and the cons.

And cutting out the carbs to treat epilepsy. I meet the parents who are using diet to help their son's seizures.

Clip

Parent

So, he's tried four different medications and none of them have helped.

Porter

Did all the medicines that he was taking, did they have an effect on him?

Parent

Yeah, he's very, very sleepy. In November we had clinic with our neurologist, he wanted to add another drug and just up, up and up his medication and I had to say no because it's not Edward anymore, he's just like a different child. So, then he said the alternative is that he could be referred to go on to a ketogenic diet.

Porter

More from Edward and his family later.

But first, we continue last week's genetics theme with a fascinating legal case that hit the headlines recently. A woman with Huntington's disease - an inherited incurable neurological condition - is suing the hospital trust that looked after her father who had it, for not warning her that she could be affected too.

The outcome of her case could profoundly change doctors' responsibilities to the families of patients with inheritable illnesses. To debate how, I am joined by GP Dr Margaret McCartney and Anna Middleton, who is a genetic counsellor and Head of Society and Ethics at the Wellcome Sanger Institute in Cambridge.

Middleton

He was explicit in telling the doctors looking after him that he didn't want his daughters to know because one of them was pregnant and he was concerned that if she found out that she was at risk of Huntington's that she would have a termination of pregnancy. It came out in the court case that he actually told other relatives but he was very explicit about not telling his daughters. And so, the daughter, who was pregnant, then went on to have her child and then she apparently started to experience symptoms herself of Huntington's and that was the point where she realised that this was an inherited condition and it was likely to have come from her father. So, she then asked the doctors caring for her father, well why didn't you forewarn me and they said - Well, we couldn't breach his confidentiality.

And so, she's now suing the NHS Trust looking after her father for her duty and her right to have this information because she said she would have ended the pregnancy.

Porter

Margaret, can I just ask you what your first impression was when you read this because it's quite a striking story?

McCartney

It is and it's a very sad story on many levels as well. I have to say my reaction when I read this was - oh no, what will happen. If there's a change and we are formally required to notify all relatives of a person who is diagnosed with having a serious genetic condition by rote. So, there are lots of rules about confidentiality and the General Medical Council are very clear about how important confidentiality is but there is also the knowledge that confidentiality is not absolute in the words of the GMC. But up until now there have been very strict rules about when it is a doctor's duty to break confidentiality. And to be honest with you, they are few and far between.

Middleton

What was fascinating for me, because I sat in the public gallery at the Court of Appeal on this, was to hear the three judges talking about the exceptional

circumstances. And they were very explicit in saying - we're not expecting all doctors in all situations to be tracking down relatives and forewarning them of information. And the GMC have got very clear guidelines on when it might be appropriate to breach somebody's confidentiality, to share information with a relative. And there are certain very specific situations. And when you're trying to protect harm in others and you can clearly show that you've weighed up the risks and the benefits of sharing that information then you can do that. And certainly, working in clinical genetics over the last 30-40 years in that practice it has happened where family information is shared with relatives, even though the initial patient is not keen on that.

But what's really striking about this particular case is it first went to court and then it was struck out by the first judge, who said - the rule of confidentiality outweighs absolutely everything else. And then it was the Court of Appeal where the three judges sat and heard the case again and said - no, actually we overturn the first judgement and we say there is reasonable grounds to challenge this. And actually, getting to that point where they were saying there was a reasonable potential duty to warn, was enough because I think up until that point people actually really were quite nervous about following the guidelines because you don't want to be sued for breaking somebody's confidentiality but you do, particularly with strongly inherited genetic conditions where a gene variant does equal disease, which isn't the case for all conditions, and where you can actually do something to prevent harm, there's quite a strong argument that you should. But what I think particularly about this case that is concerning is that although there is an existence of a duty to warn, in certain situations, that isn't a legal duty and I think turning that into law is something quite different.

Porter

To be clear, at the moment, we have professional guidance on when we should think about this but we're not under a legal obligation to do so but if this woman was to win her appeal case that would go into law, become law?

Middleton

Yes, the duty to warn would become a legal duty and I think that's really contentious.

Porter

I can see the look on Margaret's face, the dread.

McCartney

Yeah, it's just one of dismay. And I think so often we see in medicine when things go from a professional duty to a legal duty what happens is enormous fear, potentially a criminal law if you don't do something, people I think become quite frightened - how far are you going to go to trace people or track people down, you know, it becomes, I think, an administrative and bureaucratic nightmare, never mind an emotional and medical one.

Middleton

And - sorry, just to chip in there - the three judges at the Appeal Court were very specific, they said we don't expect doctors to go tracking down people and we also see that this is a very exceptional situation. But the nuance of that - that's the worry that the nuance of that might be lost if it then went to a legal duty.

McCartney

Yeah and I think we've seen that so often, once difficult professional decisions start to get put into a legal framework. The breaking of confidentiality is such a rare thing for a doctor to do, you know, it's a seminal event in your career. I'm sure there are doctors who will recall the time they had to break confidentiality, for example, after they'd advised someone who was unsafe to drive not to drive, for example, something that you're legally bound to do, you must protect the public. I think if a doctor had to do that you would remember that in your career as something that was very difficult, something that caused you sleepless nights, I'm sure, and was hard to make a judgement on, even when it's clear because it's seen as such an enormous thing for a doctor to do.

Porter

Anna, to be fair, I would imagine that in the vast majority of cases like this the family would have been told and that the individual concerned, the father in this case, would want the family informed.

Middleton

Oh, absolutely and practice over the last 30, 40 years in clinical genetics have shown that is the case. And it's very, very unusual where people are absolutely adamant that they don't want their relatives to know. And even when people are unsure about contacting their relatives a conversation with a genetic counsellor to have support in doing that is often what happens and then people feel that they then can. But it is a very tricky thing to do, families are often very disparate and spread apart and you know how do you actually do it - do you email those cousins that you haven't spoken to in 20 years? I mean what words do you use? How do you frame it? I mean these are tricky conversations.

McCartney

The problem with this is that it has potentially enormous ramifications. Instead of assuming confidentiality, we might have to start assuming that people should know unless there's a reason otherwise, for example. And I think it's going to be hard to decide what genetic conditions are such that are of a severe enough nature that relatives must be informed by rote because many genetics conditions are mixed or not always fully the expressed, for example. I think it's going to be very difficult and will create a huge amount of grey areas that will cause ethical difficulty, not just for doctors but for patients and families.

Porter

Anna, one thing for sure, I mean we're right at the beginning of the gene revolution, I mean this is likely to become more of an issue rather than less.

Middleton

It's definitely going to become more of an issue and that's simply because genetic testing itself has just gone mainstream. So, historically you could only order a genetic test through specialist clinical genetic services and now the Genomic Medicine Service has been announced with plans to revolutionise genomics across the whole of the NHS where anybody can order a genetic test pretty much. And genomic testing is relevant in ENT, dermatology, obstetrics, foetal medicine - it's just gone everywhere. And so, the issues about an individual's genetic test results and what that means for them and their family are going to become part of conversations in clinical practice across the whole of the NHS.

And this raises issues about whose information is this. Genetic information, we've often felt and seen, as family information, certainly from the specialist clinical genetic services, we see it as belonging to the family as opposed to the individual. Whereas that shift in thinking is not there yet across the whole of the NHS. So, there's quite an urgent need to upskill healthcare professionals everywhere really on the implications of this sort of testing, the uncertainty that goes with it and some of the ethical issues raised. Even if you are not having a genetic test yourself in the NHS at the moment, somebody that you're biologically related to might be or they might be having a direct to consumer ancestry test or a 23andme test or some of the direct to consumer testing that you can get online. And so, it's kind of relevant to all of us whether we're actually patients at the moment or not or whether we're just patients in waiting or whether we're just citizens in society.

Porter

Anna Middleton. And there is more information on the case on the Inside Health page of the Radio 4 website.

Now to another story that has been in the news:

Clip

A nagging sore throat may be an early sign of cancer.

Porter

Just one of a number of similar headlines generated by new research into cancer of the voice box. And it piqued our interest because, judging by my experience anyway, they might have worried quite a few people.

The study was led by Willie Hamilton, Professor of Primary Care Diagnostics at the University of Exeter, and earlier I spoke to him via our Exeter studio. Apologies for the sound quality, it wasn't a great line and Willie has to use a mobile phone to link to his cochlear implant so that he can hear me. So, was he happy with the coverage in the media?

Hamilton

It was obviously hyped up quite a lot and the mainstream media picked up a lot of the sore throat issue and arguably didn't pick up the truth behind that. So, yes, it was hyped up a bit.

Porter

Well, we've got you here now, so, you tell us the truth. What exactly did you find?

Hamilton

We found out which symptoms are important for laryngeal cancer in primary care. We confirmed that hoarseness mattered. We all get hoarse from time to time with germs and many of our patients just suffer this at home, take paracetamol or whatever they want to take and get better, without doctors getting involved. But it's hoarseness that persists. Secondly, we found other symptoms that wouldn't normally have been considered to be laryngeal cancer. The one that got a lot of publicity was sore throat. But it has to be remembered that this was sore throat in the over 60s and persistent sore throat, that patients were reporting to the GP a couple of times. So, one of the findings we found, which hasn't been publicised, was that sore throat on its own only had a chance of one in 700 of being laryngeal cancer. But persistent sore throat with a patient aged over 60, returned to their GP and had other symptoms like ear ache or difficulty swallowing, those were the patients at risk. And at considerable risk - their risks were up in the 5% range.

Porter

So, they had a one in 20 chance or thereabouts of having an underlying cancer of the larynx and that's above the threshold that we would refer someone urgently to be seen under the two-week rule?

Hamilton

And the good news of course is the ENT surgeon has an easy test if he thinks laryngeal cancer is a possibility. Laryngoscope, which is a flexible small tube, which can be passed through the nose and down to look at the voice box and is an

easy quick way of looking at the larynx. We GPs don't have that equipment, so it requires referral.

Porter

Willie, let's just set the scene a little bit. Tell me what we know about laryngeal cancer in terms of how common it is, who gets it and what the current cardinal symptoms are.

Hamilton

Well it's rare. There's just under eight in a hundred new cases a year. Ten times smaller than colorectal cancer. Eighty per cent roughly were men, more common in smokers. And you ask me what do we know about the symptoms, the answer is we didn't know much. I was involved in helping to formulate the NICE guidelines in 2015 and we had no evidence from primary care about which symptoms mattered. We assumed some of the symptoms reported from specialists would be right, so we've said hoarseness, also said lump in the neck. Oddly our results showed that the second one of those to be wrong.

Porter

And for somebody listening who's had a sore throat for a week or two and they're in their 30s, they're a non-smoker and they're otherwise healthy, this research really doesn't have any relevance to them?

Hamilton

Yeah, they treat themselves. I've got a sore throat today, Mark, I've had it for two weeks and I'm not seeing my GP.

Porter

Willie Hamilton. And there is an overview of the Exeter study findings on our website.

Low carb diets are very much en vogue at the moment, mainly among people keen to shift excess weight, but there is another use that has had nowhere near as much coverage and it's well established. Very low carbohydrate - or ketogenic diets - can help people with epilepsy.

Toddler Edward Harding had his first seizure when he was baby, a moment his parents, Sky and Peter, will never forget.

Peter

The day was like there was nothing to provoke it, just a normal day - went shopping, we just had dinner, sat him down watching the TV and then he leant to his left-hand-side, just started having a seizure.

Porter

Scary.

Peter

Yeah, it was really scary. He's never done it before, so I didn't know what to do. So, I got him out and then rang an ambulance straightaway. He stopped by the time the paramedics got there. Just as they was putting him in the ambulance he went into another seizure as well for another 10 minutes.

Sky

Peter rang me, so I came to the hospital.

Porter

In quite a state I would imagine.

Sky

Yeah very upset. He was in resus, I was being sick because I was so scared because I'd never seen it before. So, we got discharged after a week, he was on antibiotics, two weeks later I woke up at half five and it sounded like he had hiccups. I actually looked at him and he was fitting. So, obviously I got him out, called an ambulance, and we got taken down to hospital. He went status for two hours.

Porter

That's continual...

Sky

Continual seizures.

Porter

...seizures, yeah.

Sky

He was pumped with five different rescue medications; the crash team were called and then he was put in the adult ICU...

Porter

Intensive Care.

Sky

...and then they finally got it under control.

Porter

Give me an idea, over the last couple of years, of how many different types of medications he's tried.

Sky

So, he's tried four different mediations and none of them have helped.

Porter

And when you say none of them have helped, that meant he's carried on having seizures...

Sky

Yeah, yeah.

Porter

...how frequently?

Sky

So, every day.

Peter

Sometimes he has absent seizures up till like 30 times a day.

Porter

So, he sort of appears to switch off for a bit.

Peter

Yeah, he just zones out for a few seconds then comes back.

Sky

And he can drop and he does hit his head and things like that.

Porter

I mean you always have to keep an eye on a toddler but a toddler like...

Sky

But you literally cannot take your eyes of him.

Porter

Did all the medicines that he was taking, did they have an effect on him?

Sky

Yeah, he's very, very sleepy. In November we had clinic with our neurologist, he wanted to add another drug and just up, up and up his medication and then I had to say no because it's not Edward anymore, he's just like a different child. So, then he said the alternative is that he could be referred to go on to a ketogenic diet.

Champion

So, my name is Helena Champion and I'm paediatric dietician working at Cambridge University Hospital.

So, we've known that altering your food, so that you make ketones, has been beneficial for seizures since the early 1920s. When you have a low carbohydrate diet your body will stop using glucose as its primary source of energy in the blood and start making alternative fuel by burning fat. So, you can either burn the fat that you eat on a daily basis or you can burn your body fat. And as you burn that fat you make ketones as a side product.

Porter

So, the basic principles are to increase ketone levels, to decrease sugar levels and what's beneficial in the ketogenic diet, do we think it's those ketones or the lack of carbohydrate?

Champion

It's probably a little bit of both and it might actually be neither of those but the other products that come along when you make ketones may also affect how seizures initiate. But we also know that ketones can act like a drug, in that they actually will change the neurotransmitters in the brain. So, another way that ketones can affect whether your seizures will start is by having a drug like effect, a physical effect, of actually stopping that neurotransmitter initiation.

Porter

Dampening down electrical activity effectively. And what about the role of the lack of sugar, because essentially all carbohydrates are broken down into sugars, could it be that the lack of sugar is important too?

Champion

Indeed, so we get a flattening of blood sugars when we go on a low carbohydrate diet and there are some theories that show that even without ketones being present you can improve seizure control.

Porter

And are we sure that this diet works well?

Champion

In 2012, the NICE guidelines recommended, as a therapy for children with complex epilepsy, that the ketogenic diet is effective. So, we talk about a complex epilepsy as a child who has failed two targeted medications to control their seizures and we should be thinking about ketogenic diet amongst a range of other therapies at that point in your journey.

Sky

So, we started the diet on December 1st, seizure wise we hardly have any absence anymore, so we maybe have like five a day maybe.

Porter

And he was having 30 of these before.

Sky

Yeah, 30, 40 a day. The tonic clonic and the focal seizures haven't gone just yet...

Porter

These are the bigger seizures, yeah.

Sky

Yeah, but I'm hoping that as time goes on that his body starts to get used to it all.

Porter

Have they explained why his epilepsy's been so hard to control?

Sky

He had a basic gene panel done, that was negative but now they've done a more detailed one.

Porter

So, at the moment they don't know but they think it could be something genetic?

Sky

Because he has developed mental delay as well, that's quite a normal link to genetics.

Porter

But you're pleased with the impact so far?

Sky

Yeah, definitely. I mean his walking's really improved, I think his speech is getting a little bit better.

Porter

Because it's quite a lot of work, isn't it, for you...

Sky

Yes it is, yeah.

Porter

...I'm going to say ketogenic diet, it sounds simple following a special diet but the principal idea is that he has little or no carbohydrate.

Sky

So, it's an individual based diet. All meal plans are done by the dietician.

Porter

And what were you told about the likely impact?

Sky

They usually say two years before you start to ween off drugs.

Porter

So, that would be the aim, hopefully, so that if he's well controlled that you might be able to reduce one of the medicines that he's taking?

Sky

Yeah, definitely.

Porter

He's a big lad though isn't it?

Sky

Yeah, very big lad, yeah, he is.

Porter

He looks healthy and strong.

Sky

Yeah, he is.

Champion

Typically, in our centre, we will have children who've tried three, four, five medications before they're referred to try ketogenic diet therapy. It's most important that with epilepsy that you do get control of the seizures and we know that for non-complex epilepsy the drugs are very, very good but there's this group of around 30% of all epilepsies that don't respond well to those targeted first therapies and so they're much more difficult to control.

Porter

What about its role earlier on? I mean parents might understandably want to try something like this earlier on, as an alternative to drugs or maybe along with the first drug?

Champion

So, there are some studies going on to see if it has the same effect if we bring it into the mix of therapies earlier on. But those studies have still not been trialled.

Porter

What sort of response do you expect to see?

Champion

What we're looking for is stopping seizures going on for more than half an hour because if you have a seizure that goes on for any length of time over the half hour then you'll get decreased oxygen levels to the brain and that can cause some permanent changes. In this group of children with complex epilepsy if we were to put in a fourth, fifth, sixth drug we would expect 5%, so one in 20 children, to get a good clinical response in their seizure control. When we put the diet in at the same point, we get around a 60-70% clinical response to putting the diet in. So, it's very effective but it's a very onerous therapy to do because it's every meal of every day with no days off. As soon as you put carbohydrate into the diet you stop making ketones and you can get some breakthrough seizures.

Porter

Is there any evidence of any harm done through the low carbohydrate side of this, the fact that they're not getting the carbohydrate and energy that they normally would or can they get it all from fat and protein?

Champion

As long as your blood sugars remain above a critical low level then there's absolutely no problem with having a low carbohydrate diet. Younger children, under the age of two, are more at risk of having low blood sugars.

Porter

How long would they be on this diet for? Is it potentially life long?

Champion

So, no, so, the recommendations are that we try the diet for 12 weeks, if you're a responder and get good control over your seizures we run the diet for two years. And then about 80% of the children at two years, when we bring them off the diet, they find they maintain their seizure burden as if they were on the diet. And about 20% their seizures are worse and they elect to go back on the diet. So, it is a therapy you can do for a longer term but we find at two years epilepsy may well have changed and the brain has become more resilient and so you don't actually need the therapy at that point. So, the diet's not a cure but it allows the brain a bit of breathing space to gain its resilience to actually manage to control the seizures itself.

Porter

What proportion of patients that you see or their families manage to maintain the diet to the required standard? It's quite a difficult diet to follow.

Champion

Yeah, we don't not have mistakes but they get back on to diet because they know their children is much better on it. And also, our teenagers know they're better on it, so they often say to us - I don't like it but I feel better on it, so I'll stick with it.

Porter

Paediatric dietitian Helena Champion talking to me at Addenbrooke's Hospital. And there are more details on the ketogenic diet on the website.

We end the programme on a lighter note - a transatlantic spat. A picture posted by Andy Murray while recovering after his hip surgery last week prompted criticisms by doctors from America that his right arm was too hairy. They think it should have been shaved before a cannula and drip were put into his veins to reduce the risk of infection. But are they right? Time to call in umpire Margaret McCartney.

Margaret, a contentious area

McCartney

There's a delicious debate about this that actually goes back decades in medicine. So, I don't know if you remember, you're probably too young Mark, but in the 1960s and '70s women were routinely told to shave their perineums or bottoms when they expected to give birth to a baby vaginally in order to make the process somehow hygienic. Absolute rubbish, there was never any evidence whatsoever that this prevented infections afterwards, which was supposedly the aim of it. So, since then, I think there has been a bit more of an interrogation of the evidence around routine pre-operative shaving or shaving before you put in cannulas or drips. Now to be fair to the US organisation, the Association for Vascular Access, they stated that Sir Andrew Murray's arm hair should be clipped rather than shaved and it's a different thing. Shaving is a more invasive process where the surface of the skin is very often damaged, whereas clipping, the idea is, doesn't damage the surface in quite the same way. But to be honest with you Mark, I still think they're wrong.

Porter

I mean the idea makes sense, doesn't it, if you're going to have something put into your arm, I mean he's got very hairy arms, you wouldn't want all the plaster stuck to all the hair, so you can see why people might do it but what's the downside?

McCartney

There's two parallel issues, I think. One is this idea that clipping is better than shaving and one is why shave or try to remove hair at all. And I think medicine's got itself in a bit of a mess in that it has traditionally viewed hair as being something a bit dirty and something a bit unhygienic, which I don't really think is

the case very often at all. Now some surgeons might say that it makes it easier to do a good job if they have a clear field, if there's no hair on a hairy person, at their site of surgery perhaps that maybe true. But the argument here is around whether or not putting in an intravenous cannula in a hairy arm is made safer, in view of infection risk, by removing some of that hair either by shaving or by clipping. Shaving, there's certainly evidence that it does harm, we don't want to do that, it seems to damage the skin surface, make it easy for bacteria to get where they shouldn't be, particularly in or near a cannula or catheter, whatever you're putting in. Clipping, it's a bit more of an ambiguous situation. There is no clear evidence that clipping makes infection less likely. The argument may be that clipping makes it easier to put dressings on to fix the intravenous cannula better in place so that it's less likely to be damaged. But there is no high-quality evidence that says that is the case.

Porter

And looking at his as a man who's also got a hairy arm, the other advantage, of course, is that when you remove the dressings, once you leave the hospital, if you've been clipped, I suppose it's much less painful.

McCartney

Well whereas they don't know that for sure, so we don't know whether there's a difference in pain whether hair has been clipped rather than left along. And there's also lots of different types of dressings and of course some dressings are more adherent than others. And certainly, there's lots and lots of gaps in the evidence about which dressing is, I suppose, less painful to take off at the end of the day and also which is less likely to cause infection while it's in.

Porter

So, if you've got to make a decision as umpire you're voting for?

McCartney

Very much with British and Scottish team here.

Porter

Thank you very much, Margaret.

Just time to tell about next week when I try a high-tech alternative to medication for troublesome migraine. And new research into haemochromatosis - a condition which can cause problems ranging from fatigue and lack of sex drive, to arthritis and liver disease. And yet it is often missed, despite being relatively easy to diagnose. Join me to find out why.

ENDS

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