



The genetics of dystonia in CRPS – not what we were expecting

AUGUST 5, 2010 BY [BIM](#)

This is yet another important study from the **TREND group** in the Netherlands. **Bob van Hilten**, one of that rare breed who is both terrifically successful and remarkably nice, a generous, physical and intellectual sequoia of a man, wrote this little blurb below. As far as my meagre resources can decipher, it provides good evidence that the most likely genes to predispose or cause dystonia in



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HELP NEEDED FOR PAIN RESEARCH SURVEY

Please consider taking this online survey to help close the gap in our understanding of pain. You are likely eligible if you over 18 and suffer from neck pain, whiplash, low back pain, fibromyalgia, rheumatoid arthritis, migraines (will take only 15-minutes).

If you have no current pain or history of persistent pain (a pain problem that lasted more than 3 months) then you are also eligible to take the survey as a control subject (takes 5-minutes only).

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CRPS, don't. This doesn't mean that there is no genetic contribution, but it does mean that the genes that underpin familial dystonia are not important in CRPS-dystonia. So, are genes important at all in CRPS-dystonia, or in CRPS without dystonia for that matter? I don't think we have really answered that question yet, although there are several papers alluding to an answer. My gut feeling is that genetics probably modulate the risk, but certain mutations are neither sufficient nor necessary for CRPS. I guess we will wait and see.

The genetics of dystonia in CRPS – not what we were expecting

Prof JJ van Hilten

Complex regional pain syndrome (CRPS) is a chronic pain disorder. Approximately 25% of the patients with CRPS develop dystonia, which is characterized by involuntary, sustained muscle contractions.¹ The pathogenesis of CRPS and its relation with dystonia remain poorly understood. There is circumstantial

survey.

REHABILITATION FOR CRPS. WHAT DOES CURRENT CLINICAL PRACTICE LOOK LIKE?

Researchers at Brunel University London and the University of Oxford are conducting a survey to try to get a sense of what therapists involved in the rehabilitation of patients with complex regional pain syndrome (CRPS) do with their patients. The need for this research is clear. All recent international clinical guidelines recommend rehabilitation therapies as the core treatment for CRPS, yet there is little clinical evidence to guide practice. If we are to develop a clear model of best practice and rigorously evaluate it then we first need a clearer sense of what current practice is.

So if you are a clinician currently working in a rehabilitation context and you are currently involved in the management of patients with CRPS (types I or II) then it would be great if you could take the time to complete our survey which can be found here:

Biccoca, Italy

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evidence to suggest that genetic factors may play a role in CRPS, especially in CRPS associated with dystonia. First, the age at onset of CRPS patients with dystonia is, on average, eleven years younger than CRPS patients without dystonia.² Generally, early-onset forms of a disease are more likely to be genetic in origin, as opposed to late-onset forms of a disease.³ To date, however, no single causative gene has been identified for CRPS. There are familial dystonia syndromes in which the causative gene has successfully been identified (so-called DYT genes). We hypothesized that these DYT genes may play a role in the susceptibility to develop dystonia in CRPS. To increase our chances of finding gene mutations, we studied those DYT genes with a known function in biological pathways that are assumed to play a role in CRPS. In 44 CRPS patients with CRPS with fixed dystonia the genes DYT1, DYT5a, DYT5b, DYT6, DYT11, DYT12, and DYT16 were investigated for causal mutations. No such mutations were identified,

[Go to the CRPS Rehabilitation survey here.](#)

CONGRATULATIONS TO DR TASHA STANTON, SOUTH AUSTRALIA'S TALL POPPY!

We are all very proud here at BiM to announce to the world that Dr Tasha Stanton has been judged South Australia's Tall Poppy for 2015.

The Tall Poppy Campaign was created in 1998 by the Australian Institute of Policy and Science (AIPS) to recognise and celebrate Australian intellectual and scientific excellence and to encourage younger Australians to follow in the footsteps of our outstanding achievers.

It has made significant achievements towards building a more publicly engaged scientific leadership in Australia. Congratulations to Tasha for what is a thoroughly deserved recognition of her outstanding contributions.



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indicating that these genes do not seem to play a major role in CRPS with fixed dystonia.

About Bob

Professor Bob van Hilten has only three degrees – medical, neurological and scientific – all from the Leiden University, the Netherlands. He is considered a world expert in movement disorders and complex regional pain syndrome, which is why he is Scientific Director of the TREND consortium in the Netherlands – a multimillion Euro multi-programme project aimed at better understanding and treatment of CRPS. He recently purchased the most sophisticated interactive whiteboard/computer screeny thingie for his office and assures me that it has nothing to do with the World Cup and that he would come to work dressed up as Robin van Percy anyway. He is on that many committees (the small, important ones – not the big fatty ones) and has published that many papers that he now no longer bothers to count them. BodyinMind



Dr Tasha Stanton posing with His Honour, Governor of South Australia

THE ART OF PAIN LECTURE SERIES COMES TO YOU VIA PODCAST!

The Art of Pain was a great success. You can listen to [Professor Michele Sterling's excellent talk on whiplash by clicking here](#)

DO YOU HAVE BACK PAIN? WE NEED YOU!

We are looking for people who have back pain that has persisted for more than 3 months. Our study in Adelaide is investigating the relationship between chronic back pain and poor sleep, and the contributions of general mood and beliefs about pain.

We are asking that you complete a questionnaire about your general health, pain and sleep characteristics.

is thrilled that he squeezed another nanosecond out of his hectic schedule to write a post. Clearly, he did not write this bio.

Reference List

[1] Fahn S (1988). Concept and classification of dystonia.

Advances in neurology, 50, 1-8

PMID: [3041755](#)

[2] van Rijn MA, Marinus J, Putter H, & van Hilten JJ (2007). Onset and progression of dystonia in complex regional pain syndrome.

Pain, 130 (3), 287-93 PMID:

[17499924](#)

[3] Schork NJ (1997). Genetics of complex disease: approaches, problems, and solutions. *American journal of respiratory and critical care medicine*, 156 (4 Pt 2) PMID:

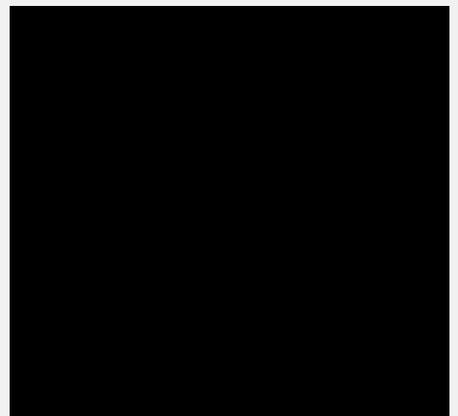
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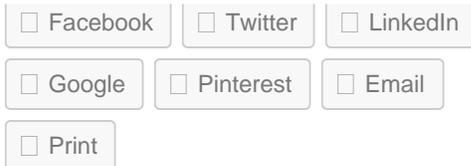
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You will also be asked to wear a wristwatch type device that records your activity levels for one week while also maintaining a pain and sleep diary. If you choose to participate, you will be given a report on your sleep quality. This research has been approved by the UniSA Ethics Committee Ref. 0000033839 “Chronic Back Pain and Sleep study”.

If you are interested, please contact Danny on 8302 1432 or email danny.camfferman@unisa.edu





Filed Under: [CRPS](#), [dystonia](#), [Guest article](#)
Tagged With: [Bob van Hilten](#), [CRPS](#), [dystonia](#)

Comments

jeisea says:

August 6, 2010 at 8:34 am

BIM you've had a couple of brilliant CRPS posts lately. I'll be posting on my blog with links to both this and the previous post. Thanks.



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3 hrs

Are you healthy, pain-free, and in Adelaide? Tory needs you for a study! It takes 1 hour and you get \$20. Please contact her at victoria.madden@mymail.unis.edu.au or 08 8302 1356 for more information.

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PARTICIPANTS NEEDED – ADELAIDE

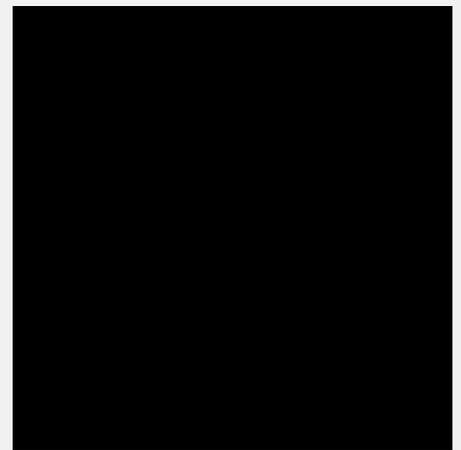
Do you live in Adelaide? Are you female between 25 and 70 and have good hearing? Do you have fibromyalgia or would you like to help someone who has by being a participant in a very

interesting and painless study we are conducting at the University of South Australia.

The study examines sensory processing in people with fibromyalgia and those without by recording your eye blink responses to some sounds. You also need to complete several questionnaires that ask about your health and well-being so we need up to 2 hours of your time.

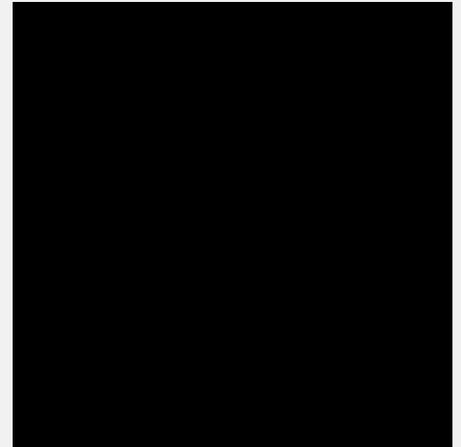
Compensation of \$20 per hour up to a maximum of 2 hours is offered. If you are interested, please contact Carolyn.berryman@unisa.edu.

BRAINMAN STOPS HIS OPIOIDS



BRAINMAN CHOOSES





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'Whiplash – to treat or reassure?'

MOTORIMPAIRMENT BLOG

- **Knee extensor power as it relates to mobility performance in people with knee osteoarthritis**

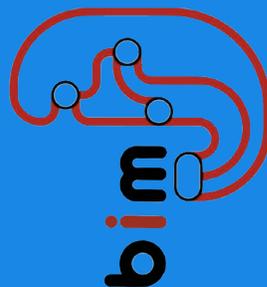
Osteoarthritis (OA) is a chronic, progressive condition characterized by a loss of articular cartilage and leads to chronic pain, disability and psychological effects in adults living with the disease. As there is no cure, researchers are concerned with identifying modifiable factors that could improve physical and psychological functioning for adults suffering with OA. The knee [...]

- **Does deep brain stimulation improve balance in people with Parkinson's disease?**

Poor balance is common in people with Parkinson's Disease (PD). This can significantly impact on quality of life. The ability to generate a step quickly and accurately after a loss of balance is critical to avoid falling. The most effective medical treatment for PD

(levodopa) seems to offer no benefit to these balance responses (King [...])

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