Apotemnophilia – the Neurological Basis of a 'Psychological' Disorder

Paul D. McGeoch[1*], David Brang[1*], Tao Song[2], Roland R. Lee[2], Mingxiong Huang[2], V. S. Ramachandran[1].

1. Center for Brain and Cognition, University of California, San Diego.
2. Department of Radiology, University of California, San Diego.

* These authors contributed equally to this work.

The question of how the human brain combines disparate sensory inputs to construct a unified body image is of longstanding interest[1, 2, 3]. We approached this subject by studying the unusual medical condition of apotemnophilia, in which otherwise mentally normal individuals express the strong and persistent desire for the amputation of a specific healthy limb[4, 5, 6]. Here we show using functional brain imaging – magnetoencephalography (MEG) – that the condition is characterised by an absence of activity in the right superior parietal lobule (SPL) when the affected limb is touched. When this discovery is combined with our earlier finding of a simultaneous increase in skin conductance response (SCR) on touching the affected limb[7], which reflects increased sympathetic nervous system activity relating to the limb[8], we conclude that what has been regarded as a purely psychological condition, actually has a neurological basis and is caused by a failure to represent one or more limbs in the right SPL. This has the bizarre consequence that although sufferers can feel the affected limb being touched, it does not actually integrate into their body image – a mismatch that results in a desire for the affected limb to be amputated.

Individuals who suffer from apotemnophilia, also termed body integrity identity disorder[5, 6], nearly always date the desire for an amputation to childhood and often struggle to explain its basis, some describing the limb as being 'over-present' and 'intrusive'. Many sufferers eventually do obtain an amputation, with most reporting feeling much happier as a result[5]. Apotemnophilia has traditionally been regarded as being strictly psychological, with explanations that range from it being a sexual paraphilia[4], perhaps related to the phallic resemblance of an amputee's stump[9], to the proposals that the mere sight of an amputee is permanently imprinted on the malleable psyche of a child as his ideal body image, or that it is simply to attract attention[6].

However, three observations suggested to us that the basis of apotemnophilia is actually neurological[10]. First, sufferers have no other psychological disturbances. Second, they desire amputation of a specific limb at a specific level. Third, there is a left-sided preponderance. Overall, we were reminded of somatoparaphrenia, a condition in which a patient with a right parietal lobe stroke denies ownership of his paralyzed left arm[2, 10]. Indeed, the right parietal lobe is known to play a vital role in constructing body image and damage to it can produce both somatoparaphrenia and various other distortions of body image, such as denial of paralysis[1, 2]. We therefore suggested that apotemnophilia also arises from dysfunction of the right parietal lobe[10]. Anatomically there is one particular area of the right parietal lobe – the SPL – that receives inputs from visual, primary somatosensory, secondary somatosensory, premotor and motor cortices[11] and thus is strategically located to combine disparate sensory inputs to construct a dynamic body image. Judging from the fact that some children with congenitally absent limbs experience phantoms[12, 13], there may be a hard-wired representation of the body in the right SPL. We postulated that if a particular limb were missing from this representation the consequence might be a desire for amputation.

Four men with apotemnophilia (AO, BC, CA, DZ) and four male controls were recruited to test our hypothesis (Table 1). Using MEG we examined right SPL activity while their feet were tapped. In each of the individual four controls tapping of either foot caused right SPL activation (Fig. 1a, b). However, in the apotemnophiles, tapping the foot that the subject desired amputated caused no right SPL activation, whereas tapping the unaffected foot (in AO, CA and DZ who desired a unilateral amputation) did cause right SPL activation (Fig. 1c, d). BC desired bilateral amputations, and stimulation to neither foot caused right SPL activation.

This confirms our hypothesis that there is a congenital failure to represent the affected limbs in their body image. Since the visual and somatosensory inputs are still intact (conveyed via intact visual and somatosensory cortices) but there is no corresponding limb representation in the right SPL, the result would be a mismatch that manifests itself as an 'intrusive' and 'over-present' limb so that the sufferer desires amputation. This interpretation is consistent with our earlier preliminary observation in two subjects (AO and BC)[7], which we have now confirmed in a third (DZ), of a significantly elevated SCR (a marker of sympathetic nervous system activity as eccrine sweat glands only receive sympathetic innervation[8]) in response to touch below the desired lines of amputation, but not above it or in the other limb[7] (Supplementary Fig. 1). Subject CA fell into the approximately 5% of individuals who show no SCR change in response to touch. We propose that this increased sympathetic response occurs because of the discrepancy between being able to feel the affected limb being touched in the absence of associated right SPL activity.

These findings fully support our hypothesis that apotemnophilia is a neurological disorder that arises from dysfunction of the right parietal lobe[10]. Conditions that stand in the borderland between neurology and psychiatry have the potential to bridge the two fields, and deliver valuable insights into how neural activity generates mental phenomena. Our findings show that a seemingly bizarre disorder, which has been long regarded as a psychological curiosity, can actually be explained in terms of specific dysfunction of known brain structures.

Methods

Ethical approval for the study was obtained and all the participants gave signed consent. The four subjects with apotemnophilia were recruited via internet support groups. The four controls were aged 27, 48, 64 and 70 years and all explicitly denied any desire for an amputation. The UCSD MEG facility has been described[14]. During the MEG session each participant underwent individual somatosensory stimulation of each foot, which was conducted at approximately two-second intervals (by PDM) using two fine, fibre-optic filaments bundled together[14]. One hundred artefact free MEG responses were averaged for each foot to increase the signal-to-noise ratio. After completion of the MEG the subjects underwent MPRAGE MRI brain scans. The MEG trial-averaged data sets were analysed using the VESTAL solution for MEG[15] for the time period between 10 and 200 ms post-stimulus, and integrated onto a FreeSurfer MRI reconstruction[16, 17]. A non-parametric permutation analysis was used to verify these activation localisations[14]. Freesurfer was used to automatically demarcate the anatomical boundaries of the right SPL and this region was then visually examined for significant MEG activations.

1. Roth, M. Disorders of the body image caused by lesions of the right parietal lobe. *Brain* **72**, 89-111 (1949)
2. Critchley, M. *The Parietal Lobes*. 1st ed. New York, United States: Hafner Publishing Company, p 225-255 (1953)
3. Smythies, J. R. The experience and description of the human body. *Brain* **76**, 132-145 (1953)
4. Money, J., Jobaris, R. & Furth, G. Apotemnophilia: two cases of self-demand amputation as a paraphilia. *J. Sex Res.* **13**, 115-125 (1977)
5. First, M. Desire for an amputation of a limb: paraphilia, psychosis, or a new type of identity disorder. *Psychol. Med.* **35**, 919-928 (2005)
6. Body Integrity Identity Disorder. www.biid.org (2009)
7. Brang, D., McGeoch, P. D. & Ramachandran, V. S. Apotemnophilia: a neurological disorder. *Neuroreport* **19**, 1305-1306 (2008)
8. Critcheley, H. D. Electrodermal responses: what happens in the brain. *Neuroscientist* **8**, 132-142 (2002)
9. Wakefield, P. L., Frank, A. & Meyers, R. W. The hobbyist: a euphemism for self-mutilation and fetishism. *Bull. Menninger. Clin.* **41**, 539-552 (1977)
10. Ramachandran, V. S. & McGeoch, P. D. Can vestibular caloric stimulation be used to treat apotemnophilia? *Med. Hypotheses* **69**, 250-252 (2007)
11. Felleman, D. J. & van Essen, D. C. Distributed hierarchical processing in the primate cerebral cortex. *Cereb. Cortex* **1**, 1-47 (1991)
12. Saadah, E. S. & Melzack, R. Phantom limb experiences in congenitally limb-deficient adults. *Cortex* **30**, 479-485 (1994)
13. Ramachandran, V. S. & Hirstein, W. The perception of phantom limbs. The DO Hebb lecture. *Brain* **121**, 1603-1630 (1998)
14. McGeoch, P. D. *et al.* Post-stroke tactile allodynia and its modulation by vestibular stimulation: a MEG case study. *Acta Neurol. Scand.* doi: 0.1111/j.1600-0404.2008.01106.x (2008)
15. Huang, M. *et al.* Vector-based spatial-temporal minimum L1-norm solution for MEG. *Neuroimage* **31**, 1025-1037 (2006)
16. Dale, A. M, Fischl, B. & Sereno, M. I. Cortical surface-based analysis: segmentation and surface reconstruction. *Neuroimage* **9**, 179-94 (1999)
17. Fischl, B. *et al*. Automatically parcellating the human cerebral cortex. Cereb. Cortex **14**, 11-22 (2004)

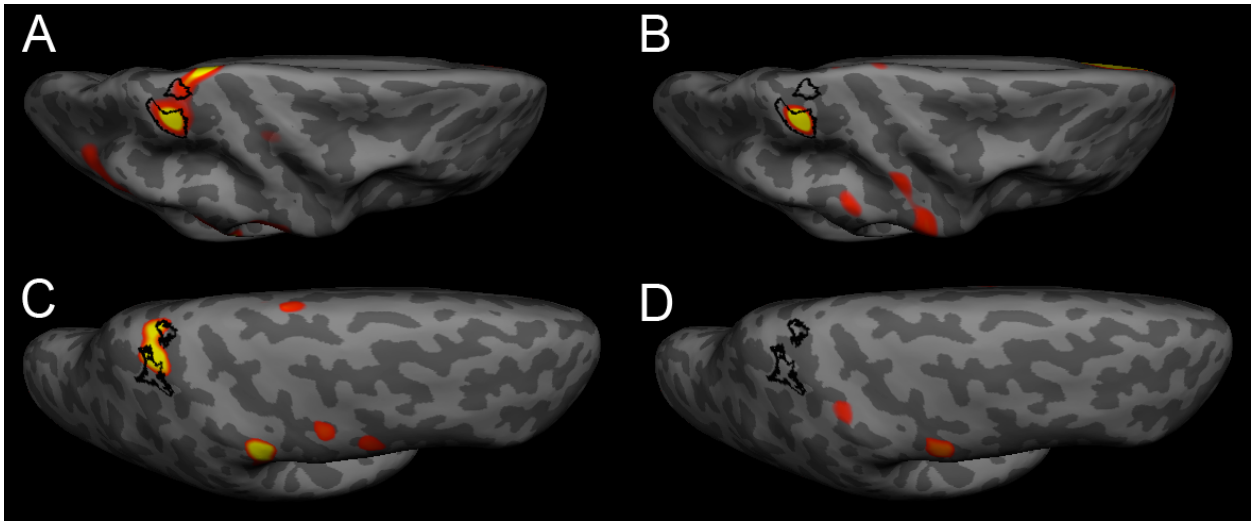

Figure 1. MEG activitions in the Right SPL

Dorsally viewed, inflated FreeSurfer reconstructions[16, 17] of the right hemisphere of an averaged control brain (a, b) and subject AO (c, d). The right SPL has been automatically outlined in black and averaged MEG activations from 60 to 120 ms analysed using VESTAL[15] are superimposed. The threshold (anything is visible) is p=0.01, the p value for the red color is p=0.001, and the saturation level (bright yellow) is p=0.0001. Images a and c show touch to the left foot of the controls and AO respectively, and b and d show touch to the right foot of the controls and AO respectively. AO was chosen as representative of all four subjects. All the normal controls independently had right SPL activation after each stimulus; they are presented in averaged form for illustrative purposes only.

SUBJECT	DESIRED AMPUTATION	FOOT	RIGHT SPL
Normal Controls	None	Left	+
		Right	+
AO (29)	Right BKA	Left	+
		Right	**-**
BC (63)	Right AKA & Left BKA	**Left**	**-**
		Right	**-**
CA (73)	Left AKA	**Left**	**-**
		Right	+
DZ (70)	Right AKA	Left	+
		Right	**-**

Table 1. Site of desired amputation and right SPL activation detected by MEG in response to tapping of the feet.

The desired site of amputation for the apotemnophiles is indicated and their age is in brackets below their initials. Whether mean MEG activation between 10 and 200 ms, using the VESTAL solution[15], reached significance in the right SPL is indicated. Supplementary Table 1 gives the actual mean MEG activations for the vertices defined as being in the right SPL. The stimulations of a limb that the subject desired to be amputated are in bold. Abbreviations: AKA = above knee amputation; BKA = below knee amputation; + = activation above level of significance; - = activation below level of significance.