The phantom and the supernumerary phantom limb: historical review and new case

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Abstract: The way we experience the world is determined by the way our brain works. The phantom limb phenomenon, which is a delusional belief of the presence of a non-existent limb, has a particular fascination in neurology. This positive phenomenon of the phantom limb raises theoretical questions about its nature. After a stroke, some patients experience the perception of an extra limb in addition to the regular set of two arms and two legs. This complex cognitive and perceptual distortion is called supernumerary phantom limb. Here, we review the pathogenesis and historical aspects, and report a new case.

Keywords: phantom limb; supernumerary phantom limb; body schema

“The phantom I had conjured up swiftly disappeared, but no spirit could have more amazed the man, so real did it seem.”

—Silas Wier Mitchell, 1872

1 Introduction

The way we experience the world is determined by the way our brain works. Compared to understanding how damage to the brain prevents some experience or behavior, it is much more difficult to explain how brain damage can create a positive symptom such as the false perception. The question of how the brain combines disparate sensory inputs to construct a unified body image has long been of interest. Under normal circumstances, we are not aware of the representation of the body in terms of differential spatial coordinate systems, but only of an integrated sense of our body in space. Corporeal awareness relies

on perceptual function and on motor programs for bodily action; it comprises a sense of the self as the object of sensory stimulation and as the agent of motor intentions and executions. Spatial perceptual disorders can arise after damage to brain areas involved in the production of the body schema. After stroke, some patients experience the perception of an extra limb in addition to the regular set of two arms and two legs: this complex cognitive and perceptual distortion is called supernumerary phantom limb (SPL)^[1] or phantom third limb^[2,3]. One case of SPL after a stroke was reported in 1930 by Ehrenwald^[4]. Although SPL may be caused by lesions in the right or left cerebral hemisphere^[5], it has been reported more frequently after right hemispheric stroke^[6].

Since 1953, there have been approximately 20 case reports on SPL^[7]. Antoniello *et al.*^[8] have suggested that stroke may result in phantom experiences more commonly than previously described. This underestimation in the past may be attributed to several factors. For example, detailed exploration into body image is not part of the standard clinical assessment. Besides, the fear of being labeled

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“crazy” for having symptoms considered unusual might discourage patients from reporting them.

2 Phantom limb: history and descriptions in popular fiction

Silas Weir Mitchell (1829–1914), one of the fathers of American Neurology, the first elected president of the American Neurological Association and a prolific writer of fiction, is usually credited with the first clinical description and the coining of the term “phantom limb (PL)”. In 1866 he published anonymously *The Case of George Dedlow*^[9] in the *Atlantic Monthly* where he described the PL syndrome in a fictional report. The central character, George Dedlow, was an army physician who was injured on the battlefield and then underwent an amputation during the Civil War. After surgery, he asked an orderly to massage his left calf, where he perceived a cramp; when the orderly drew back the cover, the amputation was revealed. In this report, PL was termed “limbs invisible”. In fact, the illusory perception of a non-existent limb in amputees was described as early as the 16th century by a French military surgeon, Ambrose Paré^[10]. In 1551, he wrote^[11]:

For the patients, long after the amputation is made, say they still feel pain in the amputated part. Of this they complain strongly, a thing worthy of wonder and almost incredible to people who have not experienced this.

Another description appeared in 1789 in a doctoral dissertation, *Dolorem Membri Amputati*, by a German physician, Aaron Lemos^[12]. In 1830, Charles Bell described in his monograph a case of a man whose arm had been amputated yet who still had perception of pain in the lost arm and of the arm changing position^[13,14]. John Hughlings Jackson, the eminent British neurologist, mentioned PL in a paper published in 1884^[15]. After Admiral Lord Horatio Nelson (1758–1805) lost his right arm at an attack on Santa Cruz de Tenerife, he experienced the common PL pain of his fingers pressing into the palm of his missing hand^[16]. Melville even mentioned PL sensations in his acclaimed novel *Moby Dick*, published in 1851^[17]. Erich Maria Remarque^[18] described in his classic novel *All Quiet*

on the Western Front the sufferings endured by the generation involved in the atrocities of trench warfare in World War I. Standing at the bedside of his severely injured friend, Franz Kemmerich, Albert Kropp asks, “How goes it Franz?” “Not so bad...but I have such a damned pain in my foot.” The leg had been amputated. The term PL reached the status of a single category in Index Medicus in 1954^[12].

3 The phenomenology of the phantom

The perception of the body may be perturbed in various ways after nervous system damage. The term PL has always been used to define erroneous perception of the persistent presence of a limb after it has been amputated. PL sensation, a component of PL-related phenomena, is only referred to as the sensation that the lost body part is still present, while PL pain includes various painful sensations in the body part that is no longer present. According to Nikolajsen and Jensen^[19], the phantom complex includes three elements: PL pain (painful sensations referred to the absent limb), PL sensation (any sensation in the absent limb, other than pain) and stump pain (pain localized in the stump). In the medical literature, SPL is the condition experienced by patients with deafferentation at some level of the central nervous system: it consists in the awareness of having an extra limb. Although phenomenologically similar, phantoms after amputation and those after a cerebral lesion differ in many aspects. For example, pain is frequently associated with PL but is only exceptionally an attribute of SPL^[20].

Recently, two phantom perceptions have been differentiated: postural and kinesthetic PL^[8]. The perception of a postural illusion is a two-step process: the realization that the limb is not in the position currently perceived and the subsequent attenuation of the phantom when the actual position of the limb is accessed by another sensory modality, most often by vision. Kinesthetic PL is often perceived as a “real” movement or an improvement of motor function. The spectrum of kinesthetic illusions ranges from simple to complex, involuntary to voluntary. One of the most astonishing features of the PL is that it may be expe-

rienced as more real than the real limb: even minor aspects of the limb are felt, such as a ring on a phantom finger^[21]. The most common PL pains are described as burning, cramping, squeezing, pins and needles, throbbing, stabbing, shocking or shooting^[22]. Henderson and Smyth, who worked with hundreds of amputees at a hospital in Germany just after World War II, observed that some of them reported that the tingling sensation of PL was pleasurable^[23]. Approximately one third of PL patients experience the phenomenon of telescoping^[24], when the distal part of the PL is gradually felt to approach the residual limb and, in the end, it may even be experienced within the stump. The sensations are neither gender- nor age-biased^[24].

Patients with SPL are fully aware of the illusory nature of their perception, which is correctly self-criticized^[25]. Hari *et al.*^[26] reported a case of a 37-year-old woman who occasionally perceived a third arm and, less frequently, a third leg, after an operation for a ruptured aneurysm, followed by an infarction of the right frontal lobe.

Phantom-related phenomena occur in at least 90% of patients with limb amputation even 25 years after the loss of the limb^[27]. Besides, phantoms are also described after anesthetic brachial plexus block^[3], spinal cord injury^[28], complete thoracic transverse myelitis^[29], peripheral nerve diseases such as polyneuritis^[30], removal of the rectum (phantom rectum)^[31] or bladder^[32], resection of hypertrophic gums^[33], penectomy^[34], and eyeball excision^[35], as well as in multiple sclerosis^[36] and in mastectomized patients^[37]. Moreover, there are also descriptions of phantom erections^[38], and menstrual cramps after hysterectomy^[39]. Phantom experiences have been described as real with a distinct shape and position also in individuals who were born limb-deficient^[40]. Poeck^[41] reported the case of a girl who was born without both forearms and hands, but incredibly, during her first year of school had learned to solve arithmetic problems by counting on her phantom fingers. Phenomenologically, the literature suggests a large spectrum of features about SPL with multimodal involvement of the tactile, visual and motor components. In many cases, the phantom is present initially for a few days or weeks, and then gradually fades from consciousness. In

others, it may persist for years, even decades^[38].

4 Case report

A 75-year-old right-handed man, a retired manual worker with a medical history of treated hypertension, was admitted to our hospital in April 2010 with sudden onset of mild weakness and sensory loss in the left arm. The patient repeatedly reported that he had a third arm protruding from his left elbow. This feeling had led him to bite the left hand to figure out who it belonged to; he was astonished by the perception of a third arm and was able to criticize it. The patient perceived the limb as if it was identical in shape to the other two arms. He felt no pain but could not control the movements of the third arm. The phantom perception was vivid, recurrent and stable. There was no history of epilepsy, psychiatric illness, or drug or alcohol misuse. Neurological examination after recovery revealed left lower VIIth nerve facial and left arm paresis, sensory loss in the left arm and the Babinski sign on the same side. At admission to hospital, the patient was well oriented with normal cognition and was able to name his body parts correctly. Assessment of the visual fields was not possible. The first cranial computerized tomography (CT) scan (April 7, 2010) revealed no evidence of a focal lesion. A second CT (April 9, 2010) showed a small area of low density in the right temporo-parietal junction. A diagnosis of stroke was suggested. Magnetic resonance imaging (MRI) confirmed the presence of an ischemic lesion. An EEG performed during the hospitalization showed mild slowing of electrical activity over the right temporo-parietal areas. Neuropsychological assessment, 15 d after the stroke, showed visual constructive and visual perceptive deficits with left neglect. The patient scored 27 out of 30 on the Mini-Mental State Examination. His belief of having a third arm had disappeared at the 2-month follow up. He has been followed for 12 months; he has retained the memory of the SPL and is amazed at what happened.

5 Pathogenesis

It is assumed that each individual develops a proprioceptive, tactile and visual image of the body. The pathogenesis of PL phenomena is still a matter of research

and remains unknown: the theories about the origins of phantom sensations can be dichotomized according to whether they focus more on peripheral or on central processes, but the mechanisms underlying these phenomena remain elusive. It has also been suggested that they may be a manifestation of a psychological disorder^[42]. Research has shown that probably both the central and the peripheral nervous systems play roles in the PL response^[43–45], with the central nervous system being a more important contributor. In 1915, the phenomenon of PL in amputees was first conceptualized as a manifestation of the persistence of the body schema for the missing limb^[46]. Since then, many theories have arisen. One of the earliest hypotheses involved neuromas: PL syndrome was attributed to ectopic irritative impulses entering the spinal cord from nervous scar tissue or neuromas in the stump of an amputated limb^[47], but the phantom can be generated centrally in the absence of nerve impulses from dermatomes associated with the phantom. In fact, in normal subjects, local anesthesia of peripheral nerves produces a phantom sensation^[48]. This does not mean that the periphery is irrelevant to all phantoms, as Carlen *et al.*^[49] asserted regarding the fact that electrical stimulation of the stump exaggerates the phantom. They proposed that the central structure that generates phantom feelings is in the spinal cord. Increased input from peripheral nociceptors has been shown to increase neuronal excitability in the dorsal horn, reduce inhibitory processes, and cause structural changes in primary sensory neurons, interneurons, and projection neurons. This is known as central sensitization and is mediated by NMDA receptors and glutamate^[50,51]. It has been suggested that PL may be due to a mismatch between efferent and proprioceptive signals, causing a moving limb to be perceived in two places^[52].

Melzack^[3] has made a significant contribution to modern hypotheses with his proposal that phantom sensations are derived from a part of the brain called “the neuromatrix”, which is a network of neurons in several brain areas and a large widespread set of neuronal connections between the thalamus and the cortex and between the cortex and the limbic system, an anatomical substrate of all the qualities of experience that are felt to originate in the body.

The neuromatrix is thought to be genetically determined but modified by experience, forming a “neurosignature” which is unique to each individual and provides each person with information about the body and its sensations. Inputs from the body may trigger or modulate the output of the networks, but are not essential for any of the qualities of experience; when an extremity is amputated, the return signal from the limb disappears, and the outgoing signals from the neuromatrix remain. This alteration of signals may be responsible for the perception of a PL^[53].

More recently, Ramachandran and Hirstein^[54] have asserted that the mechanism of phantom experiences is the “remapping” of specific brain areas. They proposed the hypothesis of topographic reorganization of somatosensory cortex in which sensation referred from neighboring receptive fields to the areas generating the phantom is one possible source for the sensation: after removal of a hand, the region of the somatosensory cortex that is de-afferented is “taken over” by afferents that normally innervate the adjacent face portion of the map. Consequently, touching the face causes referred sensations in the phantom hand. They suggested that the phenomenon results from the integration of experiences from five sources: scar tissue or neuromas in the stump, central neuronal plasticity causing the remapping of referred sensations, “the monitoring of corollary discharge from motor commands to the limbs”, a genetically-determined internal image of the body, and the presence of somatic memories of the original limb whether positional, painful or otherwise.

Mirror neurons were initially identified by Rizzolatti and his colleagues^[55] in the ventral premotor cortex in the macaque monkey. These neurons have an integral role in the observation and understanding of goal-directed actions between an agent and an object^[56]: they would allow the observer’s brain to experience the world from the other point of view. In humans, they have even evolved the ability to let a person view himself from another point of view, providing a partial explanation for introspective self-awareness^[57]. Amputees with phantom sensations may provide evidence that efficient action understanding requires a mirror neuron system that retains the representation of

an intact and functioning body. Mirror neuron activity may thus both require and reinforce the representation of the body and its functions within the body schema, even if a limb has been amputated, in which case activation of mirror neurons may reinforce the representation of the PL^[58].

Of special interest for the comprehension of the brain mechanisms underlying SPL are case studies using functional MRI (fMRI). By using fMRI in three amputees who were able to move their PL at will, Roux *et al.*^[59] showed that cortical areas devoted to the missing limb seemed to persist for several years after amputation. They further studied ten amputees by fMRI and positron emission tomography and demonstrated that primary sensorimotor cortical areas are activated by PL movements. Moreover, the activation of these areas is comparable to that produced by movements in control subjects or by movements of the contralateral normal limbs of the amputees. More recently, Staub *et al.*^[60], using a specific cognitive and fMRI protocol, investigated the clinical and anatomic correlates of chronic SPL, which developed only in association with motor intent directed at a hemiplegic-anesthetic upper limb. They reported that the SPL's movements were associated with increased fMRI responses in motor areas and in the bilateral basal ganglia-thalamus, suggesting an abnormal reorganization within the motor system.

6 Conclusion

The PL phenomenon is a delusional belief of a non-existent limb and has a particular fascination in neurology. Although it occurs in the vast majority of patients with limb amputations, clinical reports of SPL after stroke are not frequently described. The positive phenomenon of PL raises theoretical question about its nature. Psychological, peripheral and central theories have been proposed to explain the hallucinatory perception, but it continues to be a difficult condition to understand.

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幻肢和余剩幻肢研究的历史性回顾及新病例报告

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摘要: 大脑通常决定人类对其周围环境的感受。幻肢是接受过截肢手术的人所产生的一种幻觉, 他们总感觉失去的肢体仍存在于躯干上。揭开幻肢的成因已成为神经病学领域的一个研究热点。此外, 一些病人中风后会感觉到除正常存在的双臂和双腿之外, 还存在额外的肢体。这种复杂的、意识和知觉上的偏差称为余剩幻肢。本文主要就幻肢和余剩幻肢的病因及对其认识的历史性发展作一综述, 并报告一例余剩幻肢患者。

关键词: 幻肢; 余剩幻肢; 身体图式