

Case Reports

Self-Injurious Behaviour in SCA17: A New Clinical Observation

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Abstract

Background: Self-injurious behaviour has historically been associated with borderline personality disorder. Nevertheless, over recent years, it has been reported in numerous neurological syndromes, especially hyperkinesias.

Case report: Two cases of SCA17 manifested self-injurious behaviour, namely repetitive scratching of the skin resulting in severe excoriations. In one of them, the abnormal behaviour was associated with the inability to resist the impulse to commit the act along with relief following the damage.

Discussion: This is the first report describing self-injurious behaviour in SCA17, but the mechanisms underlying it are still not clear. Further studies are needed to clarify the pathophysiology of such manifestation in hyperkinetic syndromes.

Keywords: SCA17, self-injury, neurotransmitters, serotonin, dopamine

Citation: Bonomo R, Latorre A, Bhatia KP. Self-Injurious Behaviour in SCA17: A New Clinical Observation. Tremor Other Hyperkinet Mov. 2019; 9. doi: 10.7916/tohm.v0.672

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Editor: Elan D. Louis, Yale University, USA

Received: April 27, 2019; **Accepted:** August 6, 2019; **Published:** September 5, 2019

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Funding: None.

Financial Disclosures: None.

Conflicts of Interest: The authors report no conflicts of interest.

Ethics Statement: This study was performed in accordance with the ethical standards detailed in the Declaration of Helsinki. The authors’ institutional ethics committee has approved this study and all patients have provided written informed consent.

Background

Self-injurious behaviour is a clinical feature widely described in patients affected by mental retardation or psychiatric illnesses, such as psychosis or borderline personality disorder.¹

Among movement disorders, self-injury has been identified mostly in hyperkinesias, like chorea–acanthocytosis in which self-mutilating orolingual-biting is a well-recognised characteristic.² Nevertheless, such phenomenon can also be observed in Lesch–Nyhan syndrome (LNS), and it has recently been described in 6-pyruvoyl-tetrahydropterin synthase deficiency.^{3,4}

Here we report, for the first time, two cases of SCA17 presenting with self-injurious behaviour as part of their clinical symptomatology. The possible pathophysiological mechanisms underlying self-injury in hyperkinesias are further discussed.

Case I

A 66-year-old right-handed lady was diagnosed with Huntington’s disease (HD)-like 4 disorder due to SCA17 mutation (43 CAG/CAA-repeats

in *TBP* gene) at the age of 54. Disease onset was characterised by gait imbalance, falls and fidgetiness. Over the years, a progressive mood decline and cognitive deterioration accompanied the motor symptomatology. Concerning her family history, three of her siblings were similarly affected by generalised involuntary movements.

On examination, she presented with motor impersistence on eye-movement assessment and tongue protrusion. Involuntary choreic movements affected the face and limbs. The gait was broad-based. Severity of ataxia was rated with the SARA scale (25/40).⁵ Gait (4/8), stance (4/6) and speech (4/6) were significantly impaired; finger-chase test, nose–finger test, fast alternating movements and heel–shin test were as well severely affected (3/4); on sitting, there were slight difficulties with intermittent sway (1/4).

Brain MRI disclosed a severe cerebellar volume loss. On the neuropsychological assessment, the most notable finding was a significant executive dysfunction. All the other domains assessed, including memory (MMSE 21/30),⁶ visual perceptual skills, reading, naming, praxis and speed of information processing, were equally impaired.

In the last 2 years, her husband reported the development of self-injurious behaviour, namely scratching of the skin resulting in severe excoriations of the chest, arms and lower limbs. She denied itchiness. According to the partner, the skin scratching seemed to be preceded by an urge of action followed by a subsequent relaxation.

Case 2

This lady noticed for the first time a turning of the legs and curling of the hands at the age of 48. She had a family history of depression, anxiety and dementia, and reported to suffer from low mood too. On examination, she presented with generalised chorea and some difficulty with tandem gait. There were skin wounds in the upper limbs caused by repetitive self-injuring acts. Nevertheless, she was not able to report whether inner pressure or relief related to the skin scratching was experienced. Attention, concentration and memory were mildly impaired. She was admitted to hospital for further investigation. Brain MRI was normal, while the genetic screening revealed an abnormally expanded 50 CAG/CAA-repeats in *TBP* gene and a diagnosis of SCA17 was then confirmed.

Discussion

SCA17 is an autosomal dominant disorder determined by an abnormal CAG/CAA repeat expansion in *TBP* gene resulting in a pathogenic polyglutamine expansion. Normal *TBP* alleles contain 25–40 repeats. Alleles with 41–49 repeat expansion have incomplete penetrance, while alleles with 50 or more repeats were reported to have a full penetrance.^{7–9} On examination, patients can present with ataxia, chorea, dystonia, parkinsonism and seldom pyramidal signs. Also psychiatric features and cognitive impairment can be part of the clinical scenario. On brain MRI, different patterns of degeneration related to motor signs can be observed.¹⁰

This is the first report of self-injurious behaviour in SCA17, presenting as repetitive skin scratching.

Self-injurious behaviour is common in different groups of patients affected by either mental retardation or psychiatric syndromes.¹ The most typical manifestation consists of cutting and burning the skin, banging the head and limbs, picking at wounds, and chewing fingers or the lips.¹ In 1991, Winchel and Stanley speculated about the possible aetiology of such manifestations, suggesting an involvement of serotonergic, dopaminergic and opiate neurotransmitter systems. Coid and colleagues had in fact reported an increase of plasmatic endogenous opiates production in patients affected by impulsive behaviour.¹¹ On the other side, Goldestein proposed a possible dysregulation of the dopaminergic system due to the stimulation of super-sensitive D1/D2 receptors as one of the mechanisms underlying self-injurious behaviour.^{12,13} This result was based on the observation of self-biting behaviour in rats undergoing chemical denervation of dopaminergic neurons with 6-hydroxydopamine when dopamine agonists were administered.¹⁴ The same model replicated on monkeys showed again self-mutilation.¹³

Despite being extensively investigated in psychiatric disorders, over the recent years there has been increasing interest in self-injury as a behaviour distinctive of hyperkinetic movement disorders. In particular, self-biting of

lips or fingers and head banging represent a hallmark clinical feature of LNS, a rare X-linked recessive metabolic disorder due to the deficiency of the enzyme hypoxanthine–guanine phosphoribosyltransferase.¹⁵

The presence of self-injury in neurological syndromes such as LNS, but also Tourette's syndrome, further supports the role of dopamine in the phenomenology of mutilation.^{13,16–19} Intriguingly, as for tics, different authors reported that patients with self-mutilating behaviour experience an urge to injure (i.e., an increased sense of tension) prior to the act, followed by a sensation of relief.^{20,21} Compulsion and urge of action can also be observed in the impulse control disorder, where stimulation of D3 receptors and the dysfunction of frontostriatal and cingulo-frontal circuits are likely to determine the clinical manifestation.²²

Other authors have highlighted the role of serotonin in self-harming, due to the fact that 5-hydroxytryptophan, a precursor of serotonin, can dramatically improve self-injury in LNS patients.^{23,24}

Findings on rat models supported a dysfunction of the monoamine systems also in SCA17,²⁵ and by analogy, we might speculate a role of it in the origin of self-injury.

Walker and colleagues have previously suggested that the manifestation of the self-excoriations in hyperkinetic syndromes is related to an obsessive–compulsive-type behavioural disorder.²⁶ However, some studies have proved that skin picking disorder, despite resembling the compulsive rituals of obsessive compulsive disorder, has different underlying mechanisms.^{27,28}

In conclusion, to our knowledge this is the first report of self-injury in SCA17. Further studies are needed to clarify the pathophysiology of such manifestations in hyperkinetic syndromes. That might be relevant in terms of treatment consideration.

Authors' Contributions

R Bonomo, A Latorre and KP Bhatia were responsible for the study concept and design. RB and KPB were responsible for the drafting of the manuscript. All authors were responsible for the critical revision of the manuscript for important intellectual content.

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