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Hereditary geniospasm in a mother and son treated with botulinum toxin injection: A case report

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Abstract

Hereditary geniospasm is a rare and benign disorder that can cause distress and social embarrassment to patients. There are only a handful of possible treatment options available. Due to the rarity of the condition, practices differ across the world and the results are varied. These include beta-blockers, benzodiazepines and anti-epileptics. These treatments can have significant side-effects when used long term. However, botulinum toxin injections have been successfully used in a handful of cases. We report a successful botulinum treatment of hereditary geniospasm in a mother and son, with the injection protocols.

Keywords

Otolaryngology, neurology, botulinium, geniospasm

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Introduction

Hereditary geniospasm is a rare disorder of movement characterised by episodes of involuntary contractions of the mentalis muscle resulting in rhythmic lip and chin movements.[1] It is an autosomal dominant condition with almost complete penetrance associated with chromosome 9q13-q21.[2] Chin trembling is generally the only neurological finding in affected families and tends to present in early childhood, with studies showing pathophysiology similar to brainstem myoclonus.[3] There are very few cases in the United Kingdom, and no current consensus on best management. The condition can be very distressing for patients, who find the chin movements conspicuous and can suffer from troublesome tongue biting.

We present a case series of two patients, a mother and son with hereditary geniospasm, and have been successfully treated with injection of botulinum toxin into the mentalis muscle.

Case

A 2-year-old boy was referred to the Otolaryngology department from the Neurology department with persistent and involuntary episodic chin trembling, which became more pronounced when he was upset or tired. The main concern was tongue biting during the night due to his chin trembling, which caused considerable bleeding and was beginning to cause scarring of the tongue. He was otherwise well with no significant past medical history and took no regular medications.

His 40-year-old mother suffered with the same symptoms of chin trembling which had been present since early childhood, although had improved slightly over time. Her symptoms were worse with activities that required concentration, such as reading on the computer. It was also noticeable when the patient was stressed. She also suffered with occasional tongue-biting when asleep. She had no other medical problems and took no regular medications.

The diagnosis was made clinically by the neurology team based on the isolated symptoms and family correlation. They were both referred to the Otolaryngology department for treatment with botulinum. Other differentials include bruxism, tics, sleep disorders, and seizures. All patients should have a neurology consultation before coming to this diagnosis. Genetic testing was not available at our centre.

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Treatment

Both patients were treated with botulinum toxin type-A '*Dysport*' 300 units per vial. The dosage used for both patients was selected according to the manufacturers' guidelines for injection into facial muscles, which was 60 to 90 units titrated to symptoms. The *Dysport* was injected intramuscularly into the mentalis muscle in four equal aliquots evenly distributed into the four quadrants of the muscle. Due to his age, the son was treated under general anaesthesia at the same time as a previously arranged non-related surgical procedure; 90 units were injected. The mother was treated in clinic without the need for anaesthetic; 60 units in total were used. The difference in the dosage was due to the availability of the botulinum on the day of the procedure.

Outcome and follow-up

At 3 months post procedure follow up, the child had no further episodes of geniospasm following treatment. His sleeping had improved, and he had had no further tongue biting. He has since been seen every 4 months. At the 1 year follow-up, the child had a small amount of trembling, but no tongue biting. He received another botulinum injection of 90 units under general anaesthesia, due to becoming more symptomatic again.

The mother reported a significant improvement in her symptoms after 4 days post-injection. She had had no further episodes of visible geniospasm 1 month after the treatment, although she could still feel some minor spasmodic movements. However, the conspicuous chin trembling and tongue biting had abated. She was asymptomatic at her 3 months post-procedure follow up. Her ongoing follow up is similar to her son's (4 monthly), and she has not yet required further treatment at 1 year. However, both are expected to require further treatment in future as the effect of the botulinum wears off. This will be based on the reoccurrence of symptoms and how it impacts on the quality of the patients' lives, as well as the patients' wishes.

Discussion

Hereditary geniospasm, also known as 'familial chin trembling', is a genetic disorder causing repetitive movement of the chin due to repeated contraction of the mentalis muscle.[4] It has an autosomal dominant inheritance pattern with high penetrance and has been linked to chromosome 9q13-q21 in a British family over four generations.[4] There are no other associated abnormalities and it is termed a benign condition.[1] Despite this, the episodes are conspicuous and can often result in tongue biting which can be very distressing for patients. The episodes typically begin in infancy and become less pronounced in later life. It can be exacerbated by stress, emotions and concentration.[5]

Due to the paucity of cases, there is no consensus on how to best manage these patients. Prophylactic medical management, including beta-blockers, benzodiazepines and anti-epileptics have been used with limited efficacy.[6–8] The long-term treatment with these drugs carries a risk of significant side effects. Injection of botulinum toxin has been shown to be safe and beneficial in the management of localised muscle spasms and tics.[9] It has been used in geniospasm with significant improvement in symptoms.[5] Botulinum toxin, a neurotoxin produced by the Gram-positive bacteria *clostridium botulinum*, acts by blocking the release of acetylcholine at the neuromuscular junction, thus preventing muscle contraction.[10] It is a low risk procedure but may need repeating as the effects wear off.

Conclusion

In the reported two cases, botulinum injections have been an effective option for the treatment of geniospasm. These injections seem to have a quick onset and last a reasonable amount of time, as indicated by the two patients. Due to the rarity of geniospasm, we would like to add our experience of botulinum toxin to the limited literature available.

Authorship

C.P. and W.J. involved in concept design, writing of manuscript, and approval of final manuscript. J.R. and A.L. involved in concept design and approval of final manuscript and critically revised the manuscript.

Declaration of conflicting interests

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Ethical approval

Our institution does not require ethical approval for reporting individual cases or case series.

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Informed consent

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