Case study

Syndrome of fixed dystonia in adolescents – Short term outcome in 4 cases

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\textbf{A B S T R A C T}

We describe the clinical features, investigations and outcome of 4 adolescents aged 13, 16, 17 and 19 years, with fixed dystonia. The diagnosis was made within 6 months of the onset of symptoms. One patient had an identifiable traumatic precipitant. All the affected extremities had pain, sudomotor and vascular changes which were consistent with complex regional pain syndrome. The extremities affected by dystonia were the foot and the hand. The dystonia spread to affect other extremities in one patient. One patient had hemifacial spasm. Examination of the central and peripheral nervous system and allied investigations failed to reveal an organic cause. Common genetic causes for dystonia were excluded. The response to physical treatments for the affected extremities, such as Botulinum Toxin and surgery was poor. In all our cases there were significant psychological and psychiatric factors. Three patients fully met the criteria for psychogenic dystonia and responded well to psychological intervention.

Fixed dystonia in adolescents is an uncommon disorder of unknown aetiology, usually presenting in girls, which can be very disabling and difficult to treat. The affected parts of the body are usually painful and show vascular changes. The condition is allied to CRPS. Treatment with multidisciplinary approach including psychological measures and physiotherapy is more likely to be successful and may prevent unnecessary physical measures.

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1. Introduction

Dystonia is defined as abnormal muscle contraction frequently holding a body part in an abnormal position, often associated with a tremor\textsuperscript{3} and may be primary (idiopathic) or secondary (symptomatic). A fixed dystonia may occur in both primary and secondary dystonias, of multiple aetiologies, but is usually a late feature. A fixed dystonia is immobile in contrast to a primary dystonia which is typically mobile.\textsuperscript{13} Fixed dystonia, as a presenting feature, has been termed post traumatic fixed dystonia, and when associated with complex regional pain syndrome (CRPS)\textsuperscript{10} has been called the causalgia–dystonia syndrome.\textsuperscript{7}

The cause of the fixed dystonia may be elicited in some cases, by appropriate clinical history, examination and

Abbreviations: BT, Botulinum Toxin A; CRPS, Complex regional pain syndrome; CT, Computed tomography; MRI, Magnetic resonance imaging; OCD, Obsessive compulsive disorder.
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investigations. Often, when no cause can be found, a psychogenic cause may be postulated.\textsuperscript{12} Whether the cause for isolated fixed dystonia, with or without a preceding injury, is of psychogenic origin, is an ongoing controversy.\textsuperscript{5,9,14,18} This controversy in relation to the possible ‘psychogenic origin’ of fixed dystonia may reflect a lack of integrated understanding between psychiatric and neurological disciplines.

The diagnostic criteria for identifying peripheral trauma-induced dystonia and/or movement disorders has been published as the following:\textsuperscript{6} Injury must be severe enough to cause local symptoms persistent for at least two weeks or requiring medical evaluation within two weeks after the peripheral injury. The onset of the movement disorder must have occurred within a few days or months (up to one year) after the injury. The onset of the movement disorder must have been anatomically related to the site of the injury.

There are critics of these criteria\textsuperscript{18} and criticism revolves around the “denominator” problem, which can be summarised as: “given the ubiquity of peripheral trauma in the general population, why do so few people develop peripheral trauma-induced dystonia?” Various authors have tried to address this question and the hypotheses suggested can be divided into two major groups. First, people who develop peripheral trauma-induced dystonia must be predisposed to the development of dystonia\textsuperscript{19} and second, effect of sensory input on reorganisation of the central nervous system at both the segmental and cortical level.

The link between dystonia and CRPS has been postulated as abnormal sensory inhibition and poor integration of sensory input at cortical level. This is thought to apply both in primary and probably secondary dystonias.\textsuperscript{1,6,7} Existing literature about fixed dystonia may not reflect an appreciation, that the importance of the involvement of emotional processes (probably mediated in chronic disease through reorganisation of limbic as well as neocortical structures) does not necessarily mean that a neurological symptom has to be seen as psychogenic.

2. Methods

We describe the clinical features, physical findings, investigations, psychological assessment, treatment and short term follow-up of 4 cases with the onset of fixed dystonia in adolescence, with a discussion of the literature regarding paediatric onset of fixed dystonia.

3. Case descriptions

3.1. Case 1

Case 1 is female who presented at the age of 19 years. There was no family history of neurological disorder and she was the product of a non consanguineous marriage. She was born prematurely, birth weight 2100 g and gestation of 33 weeks. She was under the supervision of the paediatric neurology service for an asymmetric spastic diplegia. She demonstrated a discreet equinus deformity on her right foot and her ankles could be dorsiflexed to the neutral position. She did not have a dystonic component to diplegia which was functionally a right sided monoplegia. She had a CT scan of her brain which was reported as being normal. She did not have an MRI of her brain. She was also an insulin dependant diabetic, who had presented at 10 years of age and was controlled on insulin. Her level of diabetic control was suboptimal and this was reflected by her consistently elevated level of glycosilated haemoglobin (HbA1c).

She also had Obsessive Compulsive Disorder (OCD) and suffered from anxiety and depression, for which she took medication paroxetine, alprazolam and diazepam. She regularly would present to the psychiatric service for control of her crisis of anxiety and depression. Psychiatric assessment occurred jointly with the physical assessments.

She presented with a painful dystonia of her right foot, where she maintained the knee in extension and the foot in equinovarus. There was no history of trauma or noxious stimuli. The position of the knee improved spontaneously over time but the foot remained fixed in the equinovarus position. At the time of presentation there were sudomotor and vasomotor changes to the foot. (Fig. 1.) Her foot position did not change with sleep.

Her right foot was treated with Botulinum Toxin A injections (BT) into the tibialis posterior. She also underwent an achilles and flexor hallucis longus tenotomy with good initial results, was ambulant on crutches as she was unable to walk independently. Her right foot remained in valgus, with some internal ischiotibial rotation which was painful and correctable partially. A second infiltration of BT was administered to the ischiotibial and lateral peroneal muscles with poor results. Three months after her second injection of BT, she had another tenotomy of the achilles and peroneus. She is now partially ambulant with the aid of a splint and remained on trihexyphenidate. Her current motor function is acceptable, as she can walk without any pain, using a pair of crutches.

3.2. Case 2

Case 2 is a female of 13 years. There was no family history of significance and she was the product of a non consanguineous marriage. She was born prematurely, birth weight 1700 g and gestation of 35 weeks. She had presented at 10 years of age and was controlled on insulin. She also had Obsessive Compulsive Disorder (OCD) and suffered from anxiety and depression, for which she took medication paroxetine, alprazolam and diazepam. She regularly would present to the psychiatric service for control of her crisis of anxiety and depression. Psychiatric assessment occurred jointly with the physical assessments.

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marriage. She had two healthy siblings. She was not on any medication. There were no difficulties with her education nor did she have any history of conduct disorders. Prior to her presentation she had changed her secondary school and friends.

At the start of her course at her new school, she had an attack of anxiety which resulted in hyperventilation and tetany of both hands. She was assessed by psychology services and with psychotherapy her posture recovered spontaneously except for the right hand which only recovered partially. She was subsequently admitted for investigations (CT, MRI of the brain and neurophysiology) which were normal. A tentative diagnosis of focal dystonia and conversion disorder was made. Four days after her initial presentation, she was reassessed by clinical psychologists and psychiatrists, who confirmed a conversion disorder.

Splints were applied to her hands and wrists but could not be tolerated. She also had physiotherapy with very little effect or improvement. The hand position evolved to a fixed posture of the left hand with flexed fingers (not involving the thumb) and flexion at the wrist. The hand remained painful on mobilisation and the skin had vasomotor and sudomotor changes. (Fig. 2a and b.) There was no improvement of her hand position with sleep.

Four months after presentation she had shown some improvement as her fixed posture had partially reduced but the skin trophic changes and pain had not resolved. She had some global muscular wasting involving all the muscle groups of the left upper limb. Rest of her examination, at the time, was normal. Further repeated electrophysiological studies were normal.

She is now under monthly follow-up. Treatment with diazepam, until then, had been ineffective. She is now able to extend her arm completely and it has normal motor function, but with some pain. She has also recently begun to have symptoms and signs of Anorexia Nervosa and has been started on treatment by child psychiatrists.

3.3. Case 3

Case 3 is a female of 17 years of age. She had a normal birth history and was the product of a non consanguineous marriage. At 3 and 6 years respectively, she had repeated torsion of the L ankle and at 11 years of age, she had her ankle fixed with a good functional effect. At 13 years, she had pain in her toes and on the dorsum of her L foot and she maintained her 2nd to 5th toes in an extended and valgus position. Her neurological examination was normal. She was diagnosed with a focal dystonia. There were no precipitants.

She received 2 infiltrations of BT, 3 months apart, in the extensor digitorum longus of the left foot, with very good effect. She was able to extend her toes completely and it was free of pain for a period of time. She had a full range of movement during sleep.

At 15 years she represented with an abnormal sensation in her extremities and pain in the right dorsolumbar area. No particular cause or precipitant was identified and she was referred for a psychiatric assessment. A diagnosis of depression and anxiety secondary to an undefined organic disorder was given.

3.4. Case 4

A 17 year old adolescent girl presented 6 weeks after a bite by a dog to her right leg. Three weeks prior to presentation she...
had a diffuse headache together with some stiffness of her neck, which was worse on movement and had lasted several hours. There was intermittent tingling in right hand and foot of 3 weeks duration with difficulty in moving neck side to side lasting few hours noted for 2 weeks.

She had normal birth and development history and was born of non consanguineous parents at term. Her parents had a divorce one year prior to the onset of symptoms and she was living with her mother. Her father had been investigated for excessive fatigability and was found to have a myopathy with cylindrical spirals.

Her neurological examination was normal except for hyperaesthesia in fingers of right hand. During this admission she developed fisting, erythema and oedema of right hand and any attempts at opening her hand was very painful. Several investigations were undertaken which included a CT brain, X ray of shoulder and ankle joint which were all normal. She underwent an autoimmune screen which was negative. Her Epstein Barr Virus IgG antibody was positive suggesting a past infection. She did have a ‘flu like illness’ approximately 2 months prior to her intial presentation. All her other investigations at the time were normal. She was felt to have Complex regional pain syndrome and a regime of regular physiotherapy and pain relief was commenced with good results.

Two months after her initial presentation she represented acutely with right hemifacial spasm and a worsened posture of right hand. Her right arm showed dystonic posturing. Her shoulder was internally rotated, her elbow was flexed and pronated, with fisting and flexion at wrist. Significant allodynia involving her right hand and forearm was noted along with erythema, oedema and prolonged capillary refill. The posture of the limb was fixed and did not vary. She was unable to use any sensory tricks in order to reduce the dystonia. (Fig. 3 a–c) The position of her elbow and wrist did not change with sleep.

She had further investigations which included a normal MRI of the brain and spine, serum copper and caeruloplasmin levels were normal and genetic testing for DYT 1 mutation was negative. The diagnosis was modified to fixed dystonia with complex regional pain syndrome type I. The Chronic Pain Team provided further management of her symptoms providing a multidisciplinary approach to treatment. Various medications for pain and dystonia were tried without consistent relief. She had a cervical sympathetic block with improvement of her pain relief but minimal functional change to her limb. She also had a psychiatric assessment and subsequent psychological treatment over a period of nine months.

There were features suggestive of a somatoform disorder as the symptoms were medically unexplained and of a duration of six months. She had a significant investment in finding a medical cure associated with a reluctance to explore psychological stressors.

She received local sympathetic blocks in right upper and lower limb (Guanethedine block to calf) along with Botox injections and plaster casting of right upper limb for four weeks. After the procedure the upper limb was straight and the finger could be opened. She however continued to have allodynia and did not use her hand for any functional purpose.

Fig. 3 – (a) Case 4: Fixed dystonia with dystonia of right upper limb. Erythema and swelling of right hand can be seen. (b) Right Upper limb immobilized in plaster cast for four weeks. (c) Few days out of plaster cast, note normal posture but oedema and skin change still persisting.
During the course of her psychiatric treatment she gradually began to engage with her upper limb and feel that it was part of her body as opposed to something foreign. She was able to make a link between her physical symptoms and her psychological state and this coincided with her regaining some function of her right arm and hand so that she was begun to hand write.

Over the ensuing few months the dystonia spread to her right foot with fixed plantar flexion, inversion and curling of toes. These worsening symptoms coincided with some service reorganisation of the Chronic Pain Service and the departure of key personnel within the team.

She subsequently went on to develop symptoms and signs of Anorexia Nervosa with a body mass index of 17 and associated morbid fear of being fat with dietary restriction. She was referred to the Adolescent Eating Disorder Service for treatment.

5. Discussion

5.1. Commentaries on the cases

Case 1. This patient had an established monoplegia but the importance of structural factors in her presentation remains unclear. The onset of her dystonia may be related to spasm with pre-existing spasticity. The CRPS diagnosis is supported by the history and the pathoplastic effect of her monoplegia may explain her presentation with dystonia. Diabetes and OCD may both be regarded as stress factors and her OCD may be a reflection of underlying brain abnormality. There may also be shared aetiological factors to explain aspects of the OCD, poor diabetic control and CRPS.

Case 2 may have presented with hyperventilation syndrome. It can be postulated that the trauma that triggered her dystonia is the experience of hyperventilation. The change of school was considered as possibly an important predisposing factor. The initial treatment was psychological with no neurological input. A more co-ordinated approach may have brought clinical benefits. The later onset of Anorexia Nervosa is of interest. Anorexia is not an unusual condition in teenage girls and its significance has to be questioned. We appreciate that this case may have two separate disorders or there may have been a common pathway.

Case 3 is an account of CRPS with dystonia with apparent predisposing physical pathology that resolved. Her later presentation is ill defined and therefore the relevance of her diagnosis of depression secondary to organic disease remains speculative.

Case 4 had a diagnosis of CRPS but initial treatment seems to have been purely physical and apparently ignored the obvious psychological stressors, this may have entrenched patient and family views about organic causation.

5.2. Definition

“Fixed dystonia is an uncommon, but severely disabling condition, that usually affects young women. The pattern of muscle groups involved varies widely, but limb onset is most frequent. While the condition remains focal in a proportion of cases, spread to other muscle groups occurs in the majority. Pain is present in most patients and the abnormal posture may be accompanied by a variety of other movement disorders, sensory disturbances and...
features of CRPS. The overall prognosis is poor, but remissions can occur either with treatment or spontaneously. There is a considerable overlap of fixed dystonia with somatoform disorders/psychogenic dystonia."16

"Fixed dystonia differs from typical dystonia in a variety of ways, including the fixed posture at rest, the distribution and age at onset, the rate of progression and spread, the lack of characteristic features such as sensory tricks and action-specificity, the presence of associated features and the lack of response to traditional treatment for dystonia."12

Despite the accepted definitions fixed dystonia is generally considered to be a clinical sign and not a syndrome or diagnosis. It can occur in the context of a well defined classical dystonia with a clear aetiology, such as dystonic cerebral palsy.

CRPS1 is a syndrome that usually develops after an initiating noxious event, is not limited to the distribution of a single peripheral nerve, and is apparently disproportionate to the inciting event. It is associated at some point with evidence of oedema, changes in skin blood flow, abnormal sudomotor activity in the region of the pain, or allodynia or hyperalgesia."15 Signs and symptoms of CRPS can be found in secondary dystonia, and may be triggered by relatively minor trauma as in the non-dystonic patient. The symptoms may fluctuate in parallel to fluctuations in dystonia.

5.3. Demographics

Our patients shared many of the features described by Schrag et al.16 who described 103 patients with fixed dystonia, their relation to CRPS, somatoform and psychogenic factors. In this paper, of the 41 prospectively studied patients, 7 had an onset in teenage years. In the retrospective group 18 patients had an onset before the age of 20. The youngest presented at 11 years of age. Overall, in the combined groups 25/103 (24%) patients presented before the age of 20 years.

Hemifacial spasm has not been described in association with fixed dystonia. In a case series of 210 patients with hemifacial spasm 2.4% were diagnosed to have psychogenic hemifacial spasm and all were women.11

5.4. Psychological features

Psychological factors were important in all our cases and this was confirmed by full neuropsychiatric assessments. All our cases were assessed with the Fahn and Williams criteria9 for psychogenic dystonia. On review of the literature we found that a majority of the patients presenting in the teenage years (i.e. in the adolescent population) seemed to have overwhelming psychological features. In the report of Schrag et al.16 of the 7 patients with onset of fixed dystonia, who were teenagers, 5 had a "probable" and 2 had "clinically definite" diagnosis for psychogenic dystonia. The comorbidities of OCD and Anorexia Nervosa have not been a consistent finding in previous studies.

CRPS and psychogenic dystonia may be the outcome of a number of aetiological factors and psychological processes, which may be relevant in the formulation of some cases of fixed dystonia. However, CRPS is generally not conceived to be a psychiatric diagnosis and is not a sub-type of somatisation disorder. Where somatoform disorder is considered the diagnosis depends upon a difficult judgement about the extent to which symptoms may be explained by the underlying condition, such as CRPS. We appreciate that the classification of the cause of the fixed dystonia and CRPS is difficult and needs a sophisticated formulation. Concepts such as 'psychogenic', 'conversion' or 'somatoform disorder' and 'somatisation' may be too narrow and unidimensional.

It is recognised that good practice in the management of CRPS involves a holistic approach and avoids making a distinction between psychogenic and organic disease. That same model of understanding may be helpful in fixed dystonia.

5.5. Treatment

Schrag et al. concluded that invasive procedures, including lumbar spinal blocks and sympathectomies should be avoided in fixed dystonia. Immobilization in a plaster cast, Tibial nerve neurolysis, should also be avoided as it may result in a deterioration in the condition. In some instances, plaster casting has been postulated to prevent secondary joint damage.16 In case 4 immobilization in plaster cast lead to cosmetic improvement without any functional improvement. Overall the supposed benefits of plaster casting have not yet been fully quantified.

Operative orthopaedic procedures should also be avoided and in the case series by Schrag et al.16 resulted in further deterioration of dystonia; even amputation due to gangrene, did not prevent the progression of the dystonia in some. Addiction to high doses of opiates, anticholinergics or baclofen, was common.

The management of this condition should therefore remain conservative and multidisciplinary, incorporating psychiatric assessments, physiotherapy, pain management techniques and psychotherapy, that is individually tailored to each patient.

Botulinum Toxin, anticholinergics, baclofen and benzodiazepines may be tried, but should only be used on a long-term basis if there is evidence of definite benefit. Previous authors have used corticosteroids in patients with pseudoinflammatory changes, or bisphosphonates in those with osteoporosis,15 or intrathecal baclofen17 in fixed dystonia.

5.6. Prognosis

All the patients described initially did not have a multidisciplinary approach to their care, and investigations and psychological assessments followed dualistic processes, but as the complexity of the problem began to emerge a much more holistic model of care was embraced. Despite this, the prognosis remains poor, although a complete or partial recovery has been previously described. The best outcome seems to be on those who undergo multidisciplinary treatments early, which incorporates rehabilitation with physiotherapy and occupational therapy, as well as psychological treatments.216 In our case series the two who showed improvements with acceptable level of functioning after BT and Psychology input were those who had early intervention.
Despite appropriate intervention none returned to baseline level of functioning. One was lost to follow-up.

6. Conclusion

We present four cases of fixed dystonia in adolescents. The clinical features described in our cases are consistent with that described in the literature. In all our cases the dystonic limb was associated with allodynia and sudomotor changes which were consistent with recognised definition of CRPS. Two had favourable outcomes (with BT and psychotherapy) and two had an unfavourable outcomes. In two of our cases there was a considerable psychological element which, when addressed, showed some improvement in the dystonia. This may therefore be consistent with a diagnosis of psychogenic dystonia, but the psychological diagnostic formulation continues to remain controversial. Hemifacial spasm in the context of fixed dystonia and complex regional pain syndrome was a feature in one patient, which has not been described before.

We feel that physical treatments should only be used with specific goals in mind (such as being able to open the hand for hygiene purposes) and in the presence of an appropriate team who can ensure appropriate follow-up. The immense difficulties encountered by these patients should be acknowledged and addressed by members of the team with expertise in dealing with psychological issues.

Fixed dystonia in adolescents present us with a very complex and difficult management problem and should prompt rapid multidisciplinary and psychological assessments. Treatment and interventions should then be planned within the multidisciplinary setting specifically to avoid unnecessary and futile interventions which may be unhelpful.

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REFERENCES