how the deep gray matter is involved in the control of movement, behavior, and sleep.

VIDEO LEGEND TO FULL VIDEO CLIP
Segment 1 (0:00 – 0:36). Case 1 demonstrates repetitive stereotypical self-injurious behavior, pushing his finger into his eye. He exhibits echolalia and palilalia. The hyperkinetic movements are flailing and stereotypical.

Segment 2 (0:37 – 1:26). After weeks of sedation and ventilation, the patient was lightened for reassessment. Stereotypical cycling movements remained. There were frequent episodes of upward eye deviation plus rabbit-like oro-buccal dyskinesias.

Segment 3 (1:27 – 1:55). As the patient improved, he began talking again, although still with echolalia and palilalia. He had on-going dyskinesias (chorea), which was thought to be partly L-dopa induced.

Segment 4 (1:56 – 2:21). Before discharge, the patient is now walking, although still with occasional extra intrusive movements. He is nearly back to his premorbid function.

VIDEO LEGEND TO EARLYVIEW CLIP
Case 1 exhibits repetitive stereotypical self-injurious behavior, pushing his finger in his eye. He has hyperkinetic movements that are flailing and stereotypical. Half way into his illness, the stereotypical cycling movements remain. There were frequent episodes of upward eye deviation plus rabbit-like oro-buccal dyskinesias.

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REFERENCES

Abnormal Movements in Rett Syndrome are Present Before the Regression Period: A Case Study

Teresa Temudo, MD,1* Patricia Maciel, PhD,2 and Jorge Sequeiros, MD, PhD3,4

1Unidade de Neuropediatrics, Serviço de Pediatria, Hospital Geral de Santo António, Porto, Portugal
2Instituto de Investigação em Ciências da Vida e da Saúde (ICVS), Escola de Ciências da Saúde, Universidade Minho, Braga, Portugal
3UnIGENe, IBMC – Instituto de Biologia Molecular e Celular, Univ. Porto, Portugal
4ICBAS, Universidade do Porto, Portugal

Abstract: The suspicion of a diagnosis of Rett syndrome (RTT) is based on clinical criteria that are often not present in the first two stages of the disease, as many of its symptoms will appear at a later age. This sometimes postpones the genetic diagnosis and an early clinical intervention. We present the case of a 19-months-old girl who came to the consultation be-

*Correspondence to: Teresa Temudo, Unidade de Neuropediatrics, Serviço de Pediatria, Hospital de Santo António, SA, Largo Abel Salazar, 4099/001 Porto, Portugal. E-mail: teresatemudo@netcabo.pt
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cause of an arrest of psychomotor development noticed 5 months earlier without change in sleep pattern, behavior, or social communication. In the observation of 1 hour videotape, she presented subtle stereotypic movements of the face and hands as well as repetitive dystonic posturing of her limbs. A genetic test confirmed the diagnosis of RTT, showing a truncating mutation in the \textit{MECP2} gene (\textit{R270X}). This case confirms that stereotypic movement anomalies, albeit infrequent and subtle, are already present before the regression stage and while maintaining prehension and that, in addition, repetitive dystonic postures may occur. Recognition of these early movement disorders will improve clinicians’ ability to perform an earlier diagnosis of RTT. © 2007 Movement Disorder Society

Key words: stereotypies; transient dystonia; paroxysmal dystonia.

Stereotypies are the most constant finding in Rett syndrome (RTT) patients in all stages of the disease. Apart from the severe and fatal forms of conatal encephalopathy in RTT males, there are no RTT patients without these abnormal movements.

As we have shown recently,\(^1\) the clinical spectrum of stereotypies in RTT is very wide, and they may have a rather variable topography. In this work, we showed also, as others have done before,\(^2\) that hand stereotypies coincided with or preceded the loss of purposeful hand movements, in most patients with \textit{MECP2} mutations. Furthermore, studies based on the observation of domestic videos of RTT girls, before the period of regression, demonstrated that other subtle stereotypies, although usually unnoticed by the patient’s family, are already present before the appearance of the exuberant hand stereotypies characteristic of the disorder.\(^2\) However, to our knowledge, these videos were never published.

Herein, we present the case of a 19-months-old girl with RTT showing very subtle stereotypies and repetitive dystonic movements, before a clear regressive period and while maintaining prehension.

CLINICAL PRESENTATION

We saw this 19-months-old girl for the first time due to an arrest of psychomotor development noticed at 15 months of age. She was the first child of a young, healthy, unrelated couple, and had a normal obstetrical and neonatal history.

At birth, her occipital frontal circumference (OFC), weight and height were at the 5th percentile, but reached the 25th percentile by the age of 6 months. Then, OFC growth slowed down, progressively, until reaching the 5th percentile at 19 months of age.

She controlled her head at 3 months, grasped an object at 4 months, and had propositive prehension when she was 5 months old, without hand preference. By 8 months of age, she reached the unsupported seated position.

When she was aged 15 months, the parents became preoccupied because she did not crawl in an usual way or pass from the seated to the supine position. They did not notice any change in her sleep pattern, behavior, or social communication.

At a first glance, she was a normal, happy, and well nourished 19-months-old girl. She had no dysmorphic features and her eye contact was normal. She was interested in toys, but had no interest in their properties: she manipulated them in a very simple and stereotyped manner, and took them systematically to the mouth. She repeatedly interrupted her playing to do repetitive, dystonic movements with her right limbs (Fig. 1; Video, Segment 1). Intermittently, she also had stereotyped facial movements, such as closing of her eyes, protrusion of her tongue or lips, and grimacing (Fig. 1).

During the observation of a one-hour videotape, she twisted two fingers of the left hand, five or six times, had two instantaneous washing-like movements of her hands (Fig. 1), clapped hands three times, and twirled the hands for several times. She also had intermittent hand-to-mouth movements (Video). She seated well, without support, but could not crawl and propelled her body in a very peculiar way (Video, Segment 1). Sometimes, she tried to crawl on the four limbs, and then adopted, on occasion, a dystonic posturing of the right hand (turning it inward) (Fig. 1).

Her neurological examination, apart from the abnormal movements described earlier, was normal, as was her general examination.

Cerebral MRI was normal, but the EEG showed bilateral centroparietal spikes. Genetic testing confirmed the diagnosis of RTT, showing a truncating mutation in the \textit{MECP2} gene (\textit{R270X}).

\begin{figure}[h]
\centering
\includegraphics[width=\linewidth]{fig1.jpg}
\caption{(A) Maintenance of good social contact; (B) eye closure stereotypie; (C) protrusion of the lips; (D) hand-washing stereotypie; (E) stereotypie with hands separated; (F–H) repetitive dystonic movements of her right limbs; (I) dystonic posture of her right hand while crawling.}
\end{figure}
She was reobserved at 22 months of age. She acquired independent gait at 20 months of age. The same hand and face stereotypes were present, and more frequent, but repetitive dystonic movements were no longer observed. Manipulation was more frequently interrupted by hand stereotypes. Social contact remained good but she showed mood disturbance with irritability and laughing spells (Video, Segment 2). Head size was now below the five percentile.

**DISCUSSION**

Clinical suspicion of a diagnosis of RTT, an X-linked disorder, is based on well-defined criteria, which, nevertheless, are often not present in the early stages of the disease. This sometimes delays a genetic diagnosis and the clinical intervention. There is growing evidence, however, that development is not normal in RTT patients since infancy. Observational studies of home videos have shown that some movement abnormalities may already be present before developmental regression occurs.

This case confirms that movement abnormalities, although infrequent, are present before the regression stage of RTT, including washing-like, twirling, clapping, or twisting hand movements, as well as repetitive closure of the eyes, grimacing, and tongue protrusion. Differential diagnosis between stereotypes and tics is sometimes difficult, mainly because both can coexist in the same patient. Stereotypes have an earlier age of onset, usually appearing before the age of 2 years, and their topography is usually more distal, tics affecting proximal muscles of the limbs and face. However, both can affect all the body parts. As recently described, stereotypes in RTT can be very pleomorphic, mainly in the first decade of life, and stereotypes other than manual ones (like lip protrusion and eye closure) behave like tics, one replacing the other, and tend to disappear with time. Furthermore, there are an overlap of symptoms between RTT and autism and in the former condition tics and stereotypes frequently coexist. We hypothesize that some of the abnormal movements named as stereotypes in RTT may correspond to tics.

Besides these stereotyped movements, we demonstrate for the first time that repetitive dystonic movements may also occur, preceding exuberant stereotypes. Dystonia is a common symptom in RTT but, usually, it presents as a fixed posture and appears after some years of the evolution of the disease. Some of the repetitive dystonic limb movements of the presented patient also resemble dystonic tics. On the other hand, the hand dystonic posture when crawling (Fig. 1), resemble those described by Willemse in the syndrome of transient idiopathic dystonia of infancy; however, in that entity abnormal movements appear at rest and cease when the infant carries out propositive movements with the affected extremity and, in addition, the hemidyssmetric character of the movements favored a symptomatic etiology.

Although transient abnormal movements in the preregression stage of RTT were also referred by other authors when analyzing domestic videos of these children, to our knowledge it is the first time that these movements are described with detail and videos and photographs published. In most cases, these abnormal movements are not noticed by the parents. Often, only a long observation of the patient by a clinician, particularly if a video review procedure is included, may allow their identification.

We believe this case description and video may contribute to the early recognition of movement disorders in RTT, and potentially allow an earlier diagnosis of this disease.

**REFERENCES**

Successful Treatment of Tremor in Wilson’s Disease by Thalamotomy: A Case Report

Pramod Kumar Pal, MD, DM,1* Sanjib Sinha, MD, DM,1 Shibu Pillai, MCh,2 Arun B. Taly, MD, DM,1 and Rojin G. Abraham, MCh2

1Department of Neurology National Institute of Mental Health and Neurosciences, Bangalore, India; 2Department of Neurosurgery, National Institute of Mental Health and Neurosciences, Bangalore, India

Abstract: Little information is available on the surgical treatment of movement disorders in Wilson’s disease. We report a successful outcome of left-sided stereotactic thalamotomy in a 30-year-old man with Wilson’s disease, who had severe postural-kinetic tremor of both hands. The improvement was bilateral. Our case illustrates that stereotactic thalamotomy may be considered as an option in treating severe tremor in selected patients of Wilson’s disease and merit further trials. © 2007 Movement Disorder Society

Key words: Wilson’s disease; thalamotomy; tremor.

The common neurologic manifestations in Wilson’s disease (WD) are dysarthria, tremor, dystonia, Parkinsonism, and gait disturbances.1,2 The characteristic “wing-beating” tremor is often resistant to decoppering therapy and antitremor drugs, leading to severe morbidity. Though the usefulness of thalamotomy and thalamic deep brain stimulation for drug-resistant essential tremor and tremor predominant Parkinson’s disease has been well established, there is lack of experience in surgical treatment of tremor associated with other neurologic disorders, in particular WD. We report a young man with WD and uncontrollable bilateral upper limb tremor, who had an excellent response to unilateral thalamotomy.

CASE REPORT

A 30-year-old man presented with disabling tremor of right hand, which was severe on action and in certain postures for the past 3 years. Six months after the onset of tremor in right hand, he developed tremor in left hand, and after another 2 years, tremor of head and slurring of speech. He had jaundice 15 years and 18 months prior to his presentation to us. For the past 15 years, he was also having behavioral disturbances in the form of irritability and outbursts of anger.

He had Kayser–Fleischer rings, reduced facial expression, dysarthria, and mild restriction of upward gaze. Examination of other cranial nerves, muscle tone and power, sensory examination, stretch reflexes, and plantar responses were normal. Other findings were dystonia of left great toe, a slightly broad-based gait, and Grade 1 postural instability on pull test. Video segment 1 shows the type and severity of tremor, which was of wing beating type. Head tremor was present most of the time. When the hands were at rest, tremor was minimally present intermittently. On attempting to raise hands, an escalating tremor appeared bilaterally resulting in wide flinging movements and the limbs needed to be restrained to prevent injury. He was unable to do finger-to-nose test or hold a glass or pen.

A diagnosis of WD was confirmed by low total serum copper (42.5 μg/dl; reference range: 70–150 μg/dl), low serum ceruloplasmin (<0.072 g/L; reference range: 0.2–0.6 g/L), and high 24 hours urinary copper (700 μg/24 hours; reference range: up to 70 μg/24 hours). Ultrasound showed diffuse heterogeneous echo pattern of