COMPARATIVE STUDY OF MANUAL STEREOTYPIES IN RETT SYNDROME AND AUTISM – MOVEMENT QUANTIFICATION

ABSTRACT
Stereotypies are defined as involuntary movements, coordinated, repetitive, rhythmic, and with no specific objective. Autism and Rett Syndrome (RTT) have significant phenotypic overlap, and both are classified as pervasive developmental disorders. Hand stereotypies are common in both pathologies. Our aim is to determine quantitatively if they are really different as they seem to be by single observation. In order to characterize quantitatively stereotypic movements we designed a study based on 2d video capture and kinematic analysis. We determine the velocity, the frequency, the displacement, the extent, the duration and the number of stereotypies in both pathologies, RTT and Autism, in a population of 9 patients 4 with RTT with MECP2 mutation and 5 with Autism. The stereotypies of Autism, are faster, more repetitive, more extent and with a higher displacement, they also last less time and occurs less often than the stereotypies of RTT.

KEYWORDS – Movement disorders, Stereotypies, Rett Syndrome, Autism, 2D Movement Video Analysis, Movement Quantification

INTRODUCTION
Stereotypies are defined as involuntary movements, coordinated, repetitive, rhythmic, and with no specific objective. They can last seconds to minutes, occur in clusters, several times a day and are associated with periods of stress, excitement, fatigue or boredom. The Diagnostic and Statistical Manual of mental Disorders criteria for stereotypies requires repetitive, nonfunctioning behaviors that are present for more than 4 weeks and that interfere with
normal activities or result in self injury (DSM-IV-TR 4th ed.).

Stereotypies are classified into two major categories according to the underlying cause. They can be primary or secondary, on the basis of the presence or absence of other development problems, respectively. Primary stereotypies can be present in children with normal development and they are divided in other three subcategories as: common behaviors (e.g., rocking, head banging, finger drumming, pencil tapping, hair twisting) and two forms with atypical or complex behaviors—head nodding and complex motor movements (e.g., hand and arm flapping/waving) (Singer, H., 2009).

Secondary stereotypies are common in children with neurobehavioral syndromes, such as the autism spectrum disorders, Rett syndrome, cognitive deficits, and sensory deprivation, neurodegenerative disorders, inborn errors of metabolism, drug-induced conditions, infection, tumor, or psychiatric conditions.

The mechanism for development of stereotypies remains unknown, but the involvement of the cortico-striatal-talamo-cortical pathways, as well the dopaminergic system seem to be implicated in the pathophysiology of the stereotypies. (Kates, W et al, 2005 and Muthugovindan, D. and Singer, H., 2009). In children with autism they may be correlated with atypical prefrontal cortical development (Johnson, M H et al, 1991), frontal lobe and cerebellar vermis volume (Militerni, R. et al, 2002). Stereotypies in Rett syndrome may relate to structural and functional abnormalities of the cortico-subcortical pathways (Carter JC et al, 2008).

Rett syndrome (RTT) is the expression of a genetically determined disease, and up to 96% of classic RTT patients have a mutation located within the Methyl-CpG Binding Protein 2 (MECP2) gene on the X chromosome (Amir RE, et al, 1999 and Girard, M et al, 2001). However, there are clinically diagnosed classical and atypical RTT cases in which no mutation was identified. MECP2 mutations are rare in other disorders (eg, autism), indicating that MECP2 is only one of many genes that could contribute to the large spectrum of neurodevelopmental disorders (Gonzales, M et al, 2010).

Autism and Rett Syndrome have significant phenotypic overlap, and both are classified as pervasive developmental disorders, but the genetic basis of autism is stills unclear and likely involves multiple genes (Gonzales, M. et al, 2010).
Rett syndrome occurs almost exclusively in girls (Amir, RE et al, 1999), males, rarely, have been reported with the Rett-like phenotype (Weaving et al., 2005).

In RTT psychomotor development (PD) is normal until 6-18 months of age, followed by a period of regression of PD. The first symptoms appear in the form of psychomotor and mental retardation and stereotyped movements of hands and other body areas associated with functional loss of use of hands, deceleration of head grow. The neurological symptoms include worsening spaticity and distonia, gait dyspraxia, scoliosis, epilepsy, disordered sleep and hyperventilation (Goldman, S and Temudo T, submitted 2011).

Manual stereotypies are a hallmark of RTT, and one of its necessary and most important diagnostic criteria (Temudo, T. et al, 2007). In most of the cases they are associated with or follow the disappearance of purposeful hand movements, although they can be present before developmental regression begins (Einspieler, C. et al, 2005 and Temudo, T. et al, 2007). Stereotypies in RTT can be in the midline, with symmetrical movements of both hands, (washing, clapping, tapping, wringing, hand mouthing), or with hands apart more frequently each hand performing a different movement such as hair pulling with one hand, the other tapping the trunk (Temudo, T et al, 2008). These repetitive almost continuos and compulsive movements disappear during sleep and may aggravate with anxiety (Temudo, T. et al, 2007). In general, mutation-positive patients had more diverse stereotypies, which usually diminished after the age of 10 (Temudo, T. et al, 2007).

Autism Spectrum Disorders are a group of conditions that are neurodevelopmental in origin and first observed in infancy (Ben-Itzchak, E. et al, 2008; and Matson, JL, et al, 2007). The diagnosis is clinical and according DSM IV, based on a behavioral disorder characterized by the inability to establish social and interpersonal relationships, difficulties in verbal and nonverbal communication, as well as patterns of behavior, interests and activities are restricted, repetitive and stereotyped.

Each child with autism has his/her own repertoire of stereotypies, which can evolve with time, and is repeated always in the same manner (Muthugovindan, D. and Singer, H., 2009). However, some primary movements (bilateral flapping or
rotating the hands, fluttering fingers in front of the face, flapping/waving arm movements, and head nodding) tend to predominate (Werry, J.S. et al, 1983). Hand/finger stereotypies and gait are especially suggestive of autism (Goldman, S et al, 2008).

The target and the velocity of a voluntary movement are impaired by the movement disorders. They influence the posture, the presence of abnormal involuntary movements and the performance of normal-appearing movements (Zinner, S. and Mink, J., 2010).

Stereotypic behaviors may be verbal or nonverbal, fine or gross motor-oriented, as well as simple or complex, and they may occur with or without objects (Cunningham, A. and Schreibman, L, 2008). Stereotyped movements with object manipulations, includes spinning or twirling items, but their manipulation is not required to be identified stereotypies as such. (Zinner, S. and Mink, J., 2010) Stereotypies typically are present before 3 years of age and they can diminish or persist into adulthood (Zinner, S. and Mink, J., 2010). Autism and low IQ have additive effects on the number and variety of stereotypies. (Goldman, S et al, 2008) Stereotypies had correlation with the severity of the autism and cognitive impairment, both indexes of the extent of underlying brain dysfunction (Goldman, S et al, 2008).

The Analysis of the human body motion is a challenging problem because it is an extremely highly flexible structure that has movements in many plans; it can twist and bend in many degrees of freedom. How to extract human motion has been an important research topic in computer vision. In addition, the variety of luminance, noise induced by devices and self-occlusions of the human body complicate the movement analysis process. To address this complex problem, some investigators used marker-based methods to evaluate human body motion for clinical diagnosis (Li, et al, 2002), however, these approaches can only work for a few simple human motions.

In this work we intend to study and compare stereotypies of children with autism and Rett syndrome. We will characterize these behaviors according to the type of motion, duration, variety, frequency and velocity. We also intend to carry out research of electroencephalography in order to identify the possible abnormal electric activity during the stereotyped movements. We will use the 2D analysis of the video images and
extract the quantification of stereotypic movements.

With the data obtained we aim to achieve a more complete and accurate knowledge of stereotypes and indicate the possibility of developing this approach for early diagnostic screening of RTT and autistic patients.

SUBJECTS AND METHODS

1) Patients

In this work we propose a study with children diagnosed with Autism and Rett syndrome. The initial plan was to include 18 children, 6 with Autism and 12 with Rett Syndrome (6 with a MECP2 mutation). However, due to the selected patients age range (6 to 16 years), only 8 patients with Rett Syndrome and 9 with Autism were studied.

Our final population is composed by 4 patients with RTT with identified MECP2 mutations, and 5 patients with Autism. The other patients were excluded due to the absent of stereotypies during record or due to occlusions of the hands, which did not allow image tracking.

All the Rett patients fulfilled the revised criteria for Rett Syndrome (Hagberg, 2002 and Neul et al 2010), and all autistic patients met DSM IV Criteria for autistic disorder. The IQ'patients was assessed using the Griffiths Scale for mental development, due to the lack, or underdevelopment of language (Griffiths, R., 1992).

The parents of all children signed an informed consent for participation in this study. The study was approved by ethical commission for health of Centro Hospitalar do Porto.

2) Methods

For each child a 30 minutes video-EEG acquisition was performed in order to capture stereotyped movements.

Landmarks were attached to the patient upper body and a video electroencephalogram (EEG) system with a commercial camera was used to synchronously register EEG and video during stereotypies. All patients with Rett Syndrome were recorded with the referred setup. Each finger and parts of the upper limb were color-coded to simplify video tracking algorithms (Li, et al., 2002), in order to enable the 2D video analysis.

Finger landmarks were elastic rings and positioned in the proximal
phalanx, near to the proximal interphalange joint.

A marker with a fixed dimension (5 x 7.6 cm) has been attached to the child's clothes in the anterior and upper chest that should be visible during the entire shooting that should be performed parallel to the camera plan in order to capture the prevailing movement of the hands.

The EEG study was done simultaneously with the video, in order to correlate EEG signals with stereotyped movements.

Autistic children did not tolerate EEG electrodes nor the referred finger markers. Nevertheless, movement analysis could be performed manually.

The videos were analyzed and the segments with stereotypies (movements of interest – MOI) were cropped.

With this process we obtained 88 MOI: 44 from 4 Rett patients and 44 from 5 autistic patients. Subsequently, the videos were processed manually using MaxTRAQ®, (Innovision Systems, USA) which allows the tracking of selected finger markers or points at the hands. The tracking target was mostly the middle finger, and sometimes to avoid occlusions, other fingers or the wrist were chosen in both hands. The resultant movement data was saved in a “c3d” file and a video with the movement tracking embedded was generated.

These files were analyzed using a custom program on Matlab® (Mathworks, USA) platform from which position in xx and yy axis and speed profiles of both hands were computed. To enable a frequency analysis, the speed data was filtered using two steps: firstly the mean of speed was subtracted from the speed vector to reduce the impact of the DC (0 Hz). Secondly the speed data was filtered with a bandpass filter with lower and higher cutoff frequencies of 0.5 Hz and 10 Hz, respectively.

For the movement displacement analysis, we calculated in each MOI the Euclidian distance between the position of the tracked point and a reference point. The results achieved were filtered using the previous described steps and a Fast Fourier Transform (FFT) was applied to obtain the frequency spectrum of stereotypy movements.

The main frequency component was compared to expected range obtained by visual inspection of the MOI.
# RESULTS

<table>
<thead>
<tr>
<th>Patient</th>
<th>Age (Year)</th>
<th>Gender</th>
<th>Mutation of MECP2</th>
<th>Mental Age (Griffiths)</th>
<th>Regression</th>
<th>Epilepsy</th>
<th>EEG</th>
</tr>
</thead>
<tbody>
<tr>
<td>P1</td>
<td>11</td>
<td>Female</td>
<td>Deletion of exon 3 and 4</td>
<td>Unknown</td>
<td>Yes</td>
<td>Yes, controled with medication</td>
<td>With paroxistic activity, but not correlated with stereotopies</td>
</tr>
<tr>
<td>P2</td>
<td>8</td>
<td>Female</td>
<td>T158N</td>
<td>Unknown</td>
<td>Yes</td>
<td>Yes, controled with medication</td>
<td>Without paroxistic activity</td>
</tr>
<tr>
<td>P3</td>
<td>11</td>
<td>Female</td>
<td>R294X</td>
<td>Unknown</td>
<td>Yes</td>
<td>Yes, controled with medication</td>
<td>With paroxistic activity, but not correlated with stereotipes</td>
</tr>
<tr>
<td>P4</td>
<td>13</td>
<td>Female</td>
<td>R133C</td>
<td>Unknown</td>
<td>Yes</td>
<td>Yes, controled with medication</td>
<td>With paroxistic activity, but not correlated with stereotipes</td>
</tr>
</tbody>
</table>

**Rett Syndrome**

Mean 10 Years and 9 Months

Median 12 Years

**Autism**

<table>
<thead>
<tr>
<th>Patient</th>
<th>Age (Year)</th>
<th>Gender</th>
<th>Mental Age</th>
<th>Regression</th>
<th>Epilepsy</th>
<th>EEG</th>
</tr>
</thead>
<tbody>
<tr>
<td>P5</td>
<td>8</td>
<td>Male</td>
<td>Unknown</td>
<td>3Y e 0M</td>
<td>Yes</td>
<td>Yes, controled with medication</td>
</tr>
<tr>
<td>P6</td>
<td>10</td>
<td>Male</td>
<td>Unknown</td>
<td>3Y 4 M</td>
<td>No</td>
<td>No</td>
</tr>
<tr>
<td>P7</td>
<td>6</td>
<td>Male</td>
<td>Unknown</td>
<td>2Y 4 M</td>
<td>No</td>
<td>No</td>
</tr>
<tr>
<td>P8</td>
<td>15</td>
<td>Male</td>
<td>Unknown</td>
<td>4Y e 1 M</td>
<td>Yes</td>
<td>No</td>
</tr>
<tr>
<td>P9</td>
<td>8</td>
<td>Male</td>
<td>No</td>
<td>2Y</td>
<td>No</td>
<td>Yes, controled with medication</td>
</tr>
</tbody>
</table>

**Table I - Characterization of Study Population**
<table>
<thead>
<tr>
<th>Type of stereotypies</th>
<th>No of Stereotypies in 30'</th>
<th>No of Different Hand Stereotypies in 30'</th>
<th>Total time of Stereotypies (s)</th>
<th>Topografy Laterality, And Complexity</th>
<th>Visual Behavior</th>
</tr>
</thead>
<tbody>
<tr>
<td>Patient 1 Interlacing fingers of one hand, hand at the mouth and cross a leg over the other</td>
<td>197</td>
<td>3</td>
<td>144,12</td>
<td>Hands separated, not symmetric, not at the midline. Complex movements</td>
<td>No</td>
</tr>
<tr>
<td>Patient 2 Washing hands and clapping</td>
<td>69</td>
<td>2</td>
<td>70,44</td>
<td>Hands together, symmetric, at the midline. Single movement</td>
<td>No</td>
</tr>
<tr>
<td>Patient 3 Clapping and rubbing</td>
<td>64</td>
<td>2</td>
<td>55,44</td>
<td>Hands together, symmetric, at the midline. Single movement</td>
<td>No</td>
</tr>
<tr>
<td>Patient 4 Hand flapping, with fingertips together</td>
<td>112</td>
<td>1</td>
<td>105,4</td>
<td>Hands separated 1 always almost stopped, not symmetric, not at the midline. Complex movements</td>
<td>No</td>
</tr>
<tr>
<td>Mean</td>
<td>111</td>
<td></td>
<td>93,85</td>
<td>Mean Time (s)</td>
<td></td>
</tr>
<tr>
<td>Median</td>
<td>58</td>
<td></td>
<td>87,92</td>
<td>Median Time (s)</td>
<td></td>
</tr>
<tr>
<td>Minimum</td>
<td>2</td>
<td></td>
<td>0,92</td>
<td>Minimum Time (s)</td>
<td></td>
</tr>
<tr>
<td>Maximum</td>
<td>31</td>
<td></td>
<td>37,56</td>
<td>Maximum Time (s)</td>
<td></td>
</tr>
</tbody>
</table>

### Autism (N=5)

<table>
<thead>
<tr>
<th>Type of stereotypies</th>
<th>No of Stereotypies in 30'</th>
<th>No of Different Hand Stereotypies in 30'</th>
<th>Total time of Stereotypies (s)</th>
<th>Topografy Laterality, And Complexity</th>
<th>Visual Behavior</th>
</tr>
</thead>
<tbody>
<tr>
<td>Patient 5 Flapping</td>
<td>33</td>
<td>1</td>
<td>13,76</td>
<td>Hands separated (++) and together, symmetric, at the midline and not at the midline. Single movements</td>
<td>No</td>
</tr>
<tr>
<td>Patient 6 Flapping and trunk balance</td>
<td>25</td>
<td>1</td>
<td>11,4</td>
<td>Hands separated, symmetric, not at the midline. Single movements</td>
<td>No</td>
</tr>
<tr>
<td>Patient 7 Turn in and out, rubbing, flapping, one hand at the head and trunk balance. Hand stereotypies with object.</td>
<td>32</td>
<td>4</td>
<td>21,64</td>
<td>Hands separated (++) and together, symmetric and not symmetric, at the midline and not at the midline. Complex movements</td>
<td>Yes</td>
</tr>
<tr>
<td>Patient 8 Oscillatory tapping, tapping, “cutting” one hand with the other, finger tapping and one hand at the head</td>
<td>115</td>
<td>6</td>
<td>84,68</td>
<td>Hands together, mostly symmetric, at the midline. Complex movements</td>
<td>No</td>
</tr>
<tr>
<td>Patient 9 Tremor like movements, tickling the toy, clapping, flapping with one hand and the other at the mouth, counting like movements with the finger and twirl the wrist and cross a leg over the other. Hand stereotypies with object.</td>
<td>83</td>
<td>7</td>
<td>51,36</td>
<td>Hands together and separated, symmetric and not symmetric, at the midline and not midline. Complex movements</td>
<td>No</td>
</tr>
<tr>
<td>Mean</td>
<td>9</td>
<td></td>
<td>42,27</td>
<td>Mean Time (s)</td>
<td></td>
</tr>
<tr>
<td>Median</td>
<td>4</td>
<td></td>
<td>21,64</td>
<td>Median Time (s)</td>
<td></td>
</tr>
<tr>
<td>Minimum</td>
<td>1</td>
<td></td>
<td>0,44</td>
<td>Minimum Time (s)</td>
<td></td>
</tr>
<tr>
<td>Maximum</td>
<td>31</td>
<td></td>
<td>23,36</td>
<td>Maximum Time (s)</td>
<td></td>
</tr>
</tbody>
</table>

**Table II – Number, duration and qualitative features of stereotypies in RTT and Autism.**
<table>
<thead>
<tr>
<th>Movement Features</th>
<th>RTT (n=4)</th>
<th>Autism (n=5)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Mean Velocity (pixel/s)</td>
<td>62,68</td>
<td>371,19</td>
</tr>
<tr>
<td>Median Velocity (pixel/s)</td>
<td>57,00</td>
<td>230,95</td>
</tr>
<tr>
<td>Maximum Velocity (pixel/s)</td>
<td>279,67</td>
<td>1797,76</td>
</tr>
<tr>
<td>Minimum Velocity (pixel/s)</td>
<td>0,37</td>
<td>4,66</td>
</tr>
<tr>
<td>Mean Frequency (Hz)</td>
<td>1,46</td>
<td>2,80</td>
</tr>
<tr>
<td>Median Frequency (Hz)</td>
<td>1,18</td>
<td>2,86</td>
</tr>
<tr>
<td>Maximum Frequency (Hz)</td>
<td>4,37</td>
<td>4,96</td>
</tr>
<tr>
<td>Minimum Frequency (Hz)</td>
<td>0,64</td>
<td>0,79</td>
</tr>
<tr>
<td>Mean Desplacement (pixel)</td>
<td>242,99</td>
<td>440,04</td>
</tr>
<tr>
<td>Median Desplacement (pixel)</td>
<td>152,99</td>
<td>287,15</td>
</tr>
<tr>
<td>Maximum Desplacement (pixel)</td>
<td>1266,74</td>
<td>1891,19</td>
</tr>
<tr>
<td>Min Displacement (pixels)</td>
<td>0,92</td>
<td>21,69</td>
</tr>
<tr>
<td>Mean Extent (pixel$^2$)</td>
<td>2727,16</td>
<td>6164,88</td>
</tr>
<tr>
<td>Median Extent (pixel$^2$)</td>
<td>607,16</td>
<td>3437,84</td>
</tr>
<tr>
<td>Maximum Extent (pixel$^2$)</td>
<td>39668,74</td>
<td>45243,98</td>
</tr>
<tr>
<td>Minimum Extent (pixel$^2$)</td>
<td>0,30</td>
<td>17,86</td>
</tr>
</tbody>
</table>

**Table III - Quantitative Features of Stereotypies**

**Graph 1** - Absolute value of the difference between the mean velocity of left and right hands
Graph 2 - Duration of moments of interest

Graph 3 - Frequency of both hands in RTT and Autism
A MOI was defined as a sequence of stereotypies done continuously with minimal interval of 3 seconds.

As we can observe in Table I all the RTT patients, were girls. The age of the children was identical in the entire sample, the mean age at RTT was 10 Years and 9 Months and the median 12 years, in Autism the mean was 9 Years and 3 Months and the median 8 years. all the patients with Autism were boys.

In our population all the girls have MECP2 mutation, which is different in all of them. The oldest girls have more stereotypies (Patient 1 has 197 and patient 4 has 112) than the
youngest (patient 2 has 64) as we can see in the Table II. If we compare patient 1 and patient 3, both with same age (11 years) but with different MECP2 mutation, Patient 1 has much more stereotypies. In Table 2 Patient 1 has more stereotypies which occurs during more time (144,12 s), than patient 3 who has only 64 stereotypies in 55,44 s. We can also observe that patient 1 has more variety of stereotypies, such interlacing fingers of one hand, hand at the mouth and cross a leg over the other, than patient 3 that only has two different types: clapping and rubbing. The topography, laterality and complexity of stereotypies are variable in the study population. In RTT patients were observed stereotypic movements with hands together, hands apart, symmetrical, and asymmetrical, simple and complex movements.

We find in our sample various types of stereotypies that are common in children with RTT such as: interlacing fingers of one hand, hand at the mouth, cross a leg over the other, washing hands, clapping and rubbing. But we also find other type which is more uncommon: hand flapping, with fingertips together. These movements were found in patient 4, who has less severe mutation (R133C). All RTT patients have epilepsy although none revealed paroxistic activity synchronous with the stereotypies on the EEGs exams.

In the Table I are exhibited the mean and the median ages of the patients with autism in the study as we can see both mean (9 years and 3 months) and median (8 years) are not very different from the same parameters found in the RTT.

Analyzing table II it is notorious the difference between the number and variety of stereotypies performed by patient 8 and by the patient 9. Curiously patient 8 has a severe development delay, his mental age is 4 years and 1 month, and his real age is 15 years. Patient 8 was the child with autism that made more stereotyped movements, and some of them were very complex and difficult to describe. However patient 9 of our study has some clinical features from RTT, such as the type of stereotypies. The research of mutation MECP2 was done in this patient and was not found, despite of the phenotypic similarity.

Patient 8 exhibited 5 types of hand stereotypies as: oscillatory tapping, tapping, "cutting" one hand with the other, finger tapping and one hand at the head. He did 115 stereotypies during 84,68 s, he performed much more stereotyped
movements and had more diversity than the others patients, except patient 9 that exhibit also a large number of stereotypies: 83 with even more different types than patient 8. Patient 9 had stereotypies that resemble RTT, tremor like movements, tickling the toy, clapping, flapping with one hand and the other at the mouth, counting like movements with the finger and twirl the wrist and cross a leg over the other. However he had stereotypies with object, as patient 7, which is characteristic of autism. Patient 7 has also other aspect characteristic from Autism he looks to his hands when is doing stereotyped movements Table II. With exception of patients 7, 8 and 9, which had complex stereotypies, all the autistic patients hand single stereotyped movements.

As we observe in Table II RTT patients had more stereotypies (mean = 111) and they occur during more time in the study 30 minutes (mean = 93,85 s), than the patient with autism (mean of stereotypies = 9 and mean time = 42,27 s), whose variety of stereotypic movements was also smaller.

In the Table III are represented the qualitative features of stereotypic movements in both pathologies. As we can se stereotypies performed by patients with Autism had more elevated values of mean velocity (371,19 pixel/s), mean frequency (1,46Hz), mean displacement (440,04 pixel) and mean extent (6164,88pixel²), than RTT patients: mean velocity (62,68 pixel/s), mean frequency (2,80 Hz), mean displacement(242,99 pixel) and mean extent (2727,16 pixel²).

Graph 1 represents the absolute value of the difference between the mean velocity of the each hand in RTT and Autism and as we can observe the values corresponding to Autism are almost always highest than those corresponding to RRT.

As we observe in graph 3 the time occupied doing a MOI was clearly highest in RTT.

The Graph 3 represents the mean frequency of both hands in RTT and Autism, it is clearly that mean frequencies of RTT are mostly between 1 and 2 Hz (median = 1,18 Hz), while Autism frequencies are more scattered and high (median = 2,86 Hz) than frequencies or RTT.

Graphs 4 and 5 represents, respectively the displacement and the extent of the hands movements during a MOI, which are both highest in Autism than in RTT. This fact is also represented in the Figure 1, the displacement, represented by the
green trace, and the extent is represented by red trace, which was represents the area where the hand movement was done (Remi et al, 2011).

**Figure 1** - Trace of the displacement and the extent (red) of the movement

**DISCUSSION**

With this study we intended to characterize and achieve a better knowledge of stereotypies. As Rett Syndrome is one of the best human models to study movement disorders (Temudo, T et al, 2008), and exist an overlap between the referred syndrome and Autism (Gonzales, M. et al, 2010), these two pathologies seemed to perform an interesting model to compare stereotypies.

As we registered at the Table I, none of the Rett patients had abnormal cerebral activity during the stereotypies. So we can predict, although the limited size of our sample that stereotypies may be not related with abnormal cerebral activity, despite of the majority of Rett patients have epilepsy. Autistic patients had less epileptic pathology associated; in our sample only one child with Autism has epilepsy diagnosed, which is controlled with medication. But as was not possible to document by EEG we cannot excluded the possibility of the occurrence of abnormal cerebral activity during stereotypies on this children.

By a simple observation, stereotypies seem to be different in Rett Syndrome and Autism, although until today mostly only descriptive studies were published.

Our aim was to study the kinematic of stereotyped movements in both pathologies in order to identify the quantitative differences between them. In order to achieve our aim we performed a detailed analysis of some movement features. So as we predicted, stereotypies in Autism have a high mean velocity (371,19 pixel/s) than stereotypies in RTT (62,68 pixel/s), this shows that stereotypic movements in Autism are faster than the RTT movements.

The mean frequency in autism (2,80 Hz) is approximately the double of the mean frequency in RTT (1,46 Hz). This demonstrates that stereotypies in Autism are more repetitive than in RTT. These values are not far from those described at only
study found in our research. In that study was analyzed the stereotypic movements of a girl with RTT and their results were not distant from ours. They found a frequency movement of 1.2 Hz (Wright, M. et al., 2003).

By a simple observation stereotypic movements of RTT seemed to be less long, less extent and last for more time. With our analysis we verify that it occurred in our population. So as we described stereotypies in Autism are faster, more repetitive, more extent and has a higher displacement, last less time and occurs less often than the stereotypies of RTT. This could be explained by the occurrence of more and more severe neurological disorders in RTT, than in autism, which affects the movement performance. This last fact together with the movement features corroborates our hypothesis.

Limitations

The capacity of 2d images to study a behavior that occurs in 3d can be questionable, but some studies approved the 2d capture, by comparing the ability of this method with the 3d images analysis and realized that simple 2d images captured with a commercial camera can be used to kinematic studies with identical accuracy (Wright, M. et al., 2003).

It was difficult to have always standardized conditions, which allows better comparison between subjects and groups. Protocols for the filming and assessment of activities have the potential to counter these difficulties and enhance the objectivity of video assessment (S. Fyfe et al., 2007). In our study we had this difficulty especially with patients with autism, so in upcoming studies will be important predict these possibilities and had protocols mores adapted.

Other important limitation was the reduced population size, which not allow us to had statistically significant results and led the study to a more observational perspective. Although the results seem to be promising an extensive study would be useful to prove our results.

CONCLUSION

These initial results provide a foundation for larger projects and the first prospective observational data, despite of these observations must be confirmed in a larger sample (and replicated by other groups). As we expected there are several differences between stereotypies of
RTT and those of Autism, whose the movement features studied characterized them as faster, more repetitive, more extent and with a higher displacement, they also last less time and occurs less often than the stereotypies of RTT.

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