Movement disorders: The border between the psychogenic and the neurological

1. Introduction
2. Cases Report
3. Conclusions
1. Introduction

Frequently many neurological disorders have been shown like psychogenic disorders (paroxysmal dystonias, frontal epilepsies, etc.).

At the moment, it is a great challenge to give a correct diagnosis, differentiating between a functional psychogenic disorders to a neurological movement disorders (1)

1. Introduction

With the new diagnostic techniques advances, we have been able to identify the neurological processes underlying these pathologies (2)...

...But there are semiological processes that generate doubts in both neurologists and psychiatrists.

One of them is *kinesigenic paroxysmal dyskinesia* (KPD).

1. Introduction

Kinesigenic paroxysmal dyskinesia (KPD).

• Paroxysmal dyskinesias are a rare group of hyperkinetic movement disorders.

• Mainly characterized by their episodic nature.

• Neurological examination may be entirely normal between the episodes.

1. Introduction

**Kinesigenic paroxysmal dyskinesia** (KPD).

The diagnosis of KPD is based on not only the symptoms but more importantly, the acts that trigger those.

Substantial progress has been made in the field of genetics and KPD.

2. Case Report

We present here two cases that share genetic diagnosis (mutation of PRRT2 gene), with different path in the diagnostic process.

One of them did not raise doubts in the specialist.

Another presented multiple diagnoses before the final diagnosis (focal epilepsy, convulsive disorder, simulation disorder).

Case 1

- Female, 18 years old. Evaluated in 2013 (at age of 15) for present repeated episodes with generalized involuntary movements of brief duration (1-2 min max), without consciousness alterations.
- Appearance at posture changing and spontaneously.
- Paresthesias on lower extremities while resting that stopped when she started walking.
- FB: mother with similar episodes between 15 and 27 years old. A third cousin with childhood epilepsy.
- Twisted involuntary hands movements since she was 5 years old.
Case 1 – Complementary examinations

Analytic: subclinical hyperthyroidism
MR: normal
EEG during consciousness: normal

Video-EEG: Brain bioelectrical activity with normal baseline during sleep and consciousness. A generalized dyskinesia/dystonia on extremities appeared while phase 2 of NREM activity, with 15 seconds of length, without cortical triggers.
Case 1 - Evolution

- Treated with **LMT 50 mg/12 hrs** with partial improvement. Presenting still 5-7 episodes a day.
- Acetazolamide was added, 1 per/24 hrs
- Monitoring with kidney ecography
- Episodes in remission
- Episodes reappear when reducing LMT dose
Case 2

- Male, 26 years old.
- PB: diagnosed with *frontal epilepsy* at age 12 in Argentine
- Medical consultation for *paroxystic episodes of brief duration* with involuntary movements in left extremities, preceded by tingling in the left leg.
- No consciousness alteration.
- Frequency: more than 5 times a day.
- **Starting at initiating movement.** Other related factors: sleep privation, stress and physical effort.
- Moderate occasional oppressive headache.
- CBZ 200: 1-1-1 as treatment for many years with few control.
- CBZ was suspended because of *cognitive complaints* and doubts about the diagnosis.
Case 2 – Complementary tests

MR (2 studies): 1 centimeter diameter arachnoid cyst in middle cranial fossa, doubtful pathological origin.

EEG during consciousness: normal. No changes between 2015 EEG and 2008 study.

Analytic: without impairments
Previous Diagnostics

*Focal epilepsy and arachnoid cyst* in Argentina at 11 years old, treated with carbamazepine, but it did not control the episodes.

A year ago, the patient was hospitalized in the psychiatric unit for *depressive symptomatology*. The treatment was suspended due to cognitive complaints and diagnostic doubts, due to a *possible simulation or conversive disorder*. 
Previous Diagnostics

Worsening of the paroxysmal episodes and the anxiety, tremor in the upper extremities began during the hospitalization.

At medical discharge, he was diagnosed with:

- Anxious Depressive Disorder
- Anankastic and Schizoid Personality Traits
- Conversive Disorder
- Simulation Disorder

The patient was treated with fluoxetine.

One year later

Neurologic unit service
## Neuropsychological profile

<table>
<thead>
<tr>
<th>Category</th>
<th>Results</th>
</tr>
</thead>
<tbody>
<tr>
<td>Attention</td>
<td>Focal attention 6 (span 7)</td>
</tr>
<tr>
<td></td>
<td>Attentional control: 3 (span 4)</td>
</tr>
<tr>
<td></td>
<td>TMT-A: 22”</td>
</tr>
<tr>
<td></td>
<td>TMT-B: 51”</td>
</tr>
<tr>
<td>Memory</td>
<td>Verbal short term: 9/12 with keys 12/12</td>
</tr>
<tr>
<td></td>
<td>Verbal long term: 11/12 with keys 12/12</td>
</tr>
<tr>
<td></td>
<td>Visual: PD=25 Pc=80</td>
</tr>
<tr>
<td>Language</td>
<td>Categorical fluency: 16</td>
</tr>
<tr>
<td></td>
<td>Semantical fluency: 25</td>
</tr>
<tr>
<td>Visuoconstruction</td>
<td>Rey figure: Pd= 34 Pc= 80</td>
</tr>
<tr>
<td>Executive function</td>
<td>Zoo: Perfil 4</td>
</tr>
<tr>
<td></td>
<td>Stroop: PD: 67  PT 72</td>
</tr>
</tbody>
</table>
Psychological profile

Anxious-Depressive Disorder. (SCID-II) (Hamilton Test)

Anankastic and Schizoid Personality traits (IPDE)
Case 2 - Evolution

- Treated with acetazolamide 250 mg/24 hs with significant improvement in the episodes
- Refuses to take CBZ
- Kidney ecografía as following
- Genetic studies are made with the result of changes in the PRRT2 gen

**Diagnosis**

**Kinesigenic paroxysmal dyskinesia (KPD)**
About KPD

Paroxysmal dyskinesia psychopathology

1. Phenomenology of the episodes:

   a. **High frequency** of dystonic symptoms in PD: deficits of inhibitory mechanisms due to abnormalities in the modulation of the exit signals of the basal ganglia.

   b. **Association** with other dystonic phenomena: writer’s cramp.

2. No alteration of the **level of consciousness**

3. **Absence** of critical anomalies during the episodes.
4. **Focal lesions** in Basal ganglia are associated with secondary PD.

5. **Absence of intercritical abnormalities** in most patients. In case of occurrence (slow waves frequencies), there is no relation with the topography of the attacks.

Clinical classification of the primary paroxysmal dyskinesia

A. Inclusion criteria: 1 + 2 a, b ó c
   1) Paroxysmal attacks of dystonia, choreas, ballismus or other combinations of them, with subit start and variable duration (seconds to hours).
   2) They are classified according to the precipitating factor:
      a) Kinesigenic paroxysmal dyskinesia (KPD): abrupt and fast movements or the will to move start the attacks.
      b) Non-kinesigenic paroxysmal dyskinesia (NKPD): attacks are started by caffeine, alcohol and other non-kinesigenic factors (stress, tiredness…)
      c) Paroxysmal dyskinesia caused by exercising (PDE): the attacks are caused by intense exercise.

B. Exclusion criteria:
   1) Symptoms caused by other neurologic condition (Secondary PD)
   2) Psychogenic symptoms
Kinesigenic paroxysmal dyskinesia (KPD)

Diagnostic criteria (Bruno, 2004):
- Movements-caused attacks
- Short duration attacks (<1 min)
- No consciousness lost or pain during the attacks
- Exclusion of other organic disorders and normal NE
- Attacks are controlled with CBZ/Fenitoine
- Starting age between 1-20 years old, without family background (Sporadic >20 a: secondary)

Associations: Seizures in childhood, febrile seizures, epilepsy, migraine, writer’s cramp, TE, blepharospasm, Torette.
# Paroxysmal Dyskinesia Genetics

<table>
<thead>
<tr>
<th>Gene</th>
<th>Location</th>
<th>Inheritance</th>
<th>Phenomenology of attacks</th>
<th>Remarks</th>
</tr>
</thead>
<tbody>
<tr>
<td>PDRT2</td>
<td>18p11.2</td>
<td>AD</td>
<td>Most frequently PKD, rarely PNKD and PED</td>
<td>Allelic with BFS, ICCA, hemiplegic migraine, and other paroxysmal disorders</td>
</tr>
<tr>
<td>MR-1</td>
<td>2q35</td>
<td>AD</td>
<td>Most frequently PNKD, rarely PED</td>
<td>One family reported</td>
</tr>
<tr>
<td>KCNMA1</td>
<td>10q22</td>
<td>AD</td>
<td>PNKD</td>
<td>One manifestation of GLUT-1 deficiency syndrome</td>
</tr>
<tr>
<td>SLC2A1</td>
<td>1p34.2</td>
<td>AD</td>
<td>Most frequently PED, rarely PNKD</td>
<td></td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Dystonia as only motor feature</th>
<th>DYT1, DYT2, DYT4, DYT6, DYT7, DYT13, DYT17, DYT21, DYT24, DYT25</th>
</tr>
</thead>
<tbody>
<tr>
<td>Dystonia combined with other movement disorders</td>
<td>Parkinsonism</td>
</tr>
<tr>
<td></td>
<td>Myoclonus</td>
</tr>
<tr>
<td>Paroxysmal dyskinesias</td>
<td>DYT8, DYT9, DYT10, DYT19, DYT20</td>
</tr>
</tbody>
</table>
Pericentromeric region of chromosome 16: common substrate of paroxysmal dyskinesias and hereditary epilepsy

PKD: Gen PRRT2

EAPM
The European Association of Psychosomatic Medicine

Hospital Universitario Infanta Sofia
SaludMadrid
Comunidad de Madrid
Kinesigenic paroxysmal dyskinesia (KPD)

- 1940, Mount and Reback: “Paroxysmal choreo-athetosis”
- 1977, Lance: “Paroxysmal exercise-induced dyskinesia, (PED)”
  - 1ª classification: PDC, PKC, PED
- 1995, Demirkiran, and Jankovic:
  - “Paroxysmal kinesigenic dyskinesia” (PKD)
  - “Paroxysmal non-kinesigenic dyskinesia” (PKND)
  - “Paroxysmal exercise-induced dyskinesia” (PED)
- 2004, Bruno: diagnostic criteria of ideopathic KPD
- Genetic advances:
  - Rainier, 2004: PED. SLC2A1
  - Suls, 2008: PKND. MR-1
  - Chen, 2011: PKD. PRRT2, Cr 16
- 2014, Erro: 500 KPD patients genetically evaluated and confirmed. revisión de más de 500 PKD confirmados genéticamente. Classification proposal of the PD.
Conclusions

1. **Difficulties to make a correct diagnosis**, and the consequences that a wrong one could affect in the **quality of life** and causing the appearance of comorbidities in the patient.

2. **The assessment** that doctors must give to the symptoms as a way of determining the final diagnosis is very important.

3. It is complicated to make a good diagnosis only based on the observation of symptoms. Therefore, it is advisable to use **complementary tests** that are sometimes helpful for the final diagnosis.
Conclusions

1. Only 33% of cases of movement disorders are correctly diagnosed, and the diagnosis must be more influenced by clinical exploration than other tests (4).

2. Thanks to research and scientific advances such as genetic study, we know that there are genetic mutations associated with movement disorders.

3. The proline-rich transmembrane protein (PRRT2) gene was recently identified using genome sequencing as the cause of autosomal dominant PKD.

Conclusions

In both cases, the final diagnosis is the same.

One of them with a family history that facilitated to the doctor the neurological diagnosis and adequate treatment, and better quality of life of the patient.

The other one with a long history of diagnoses that have influenced Psychiatric comorbidity of the patient, such as anxiety, emotional regulation and social skills that generate confusion in the diagnosis.
Conclusions

The psychiatric assessment may be a guide in the evaluation of comorbidities that define the Diagnosis.

It is very important to train doctors in psychosomatic diseases that can be generated by an incorrect diagnosis.
Thank you
Guadalupe Chiclana Actis Ps.Msc.
gchiclana@excetalent.com