A NEW HISTOPATHOLOGICAL CLASSIFICATION OF MYOCLONUS EPILEPSY

SHIGEMI ANRAKU

Institute of Brain Diseases, Kurume University School of Medicine, Kurume, 830, Japan

(Received for publication September 14, 1976)

At present, histopathologically myoclonus epilepsy syndrome has been divided into 3 types, i.e. Lafora-body type, degenerative type and lipidosis type (Harriman et al., 1955; Seitelberger et al., 1964).

On the basis of the findings of 12 autopsy cases, the author classify as follows (Table 1.) (Anraku, 1974).

1. Lafora-body type (case 1 - case 4): The main histological findings were the appearance of Lafora-bodies (basophilic inclusions) first described Lafora et al. (1911). These bodies were noted all over the brain, especially they were abundant in the nerve cells of the substantia nigra, the dentate nucleus and the thalamus. Histomorphologically these appeared as rounded bodies of varying size lying singly or in groups in the neuronal cytoplasm. Moreover, in the liver cells (case 1, 3) and heart muscle

<table>
<thead>
<tr>
<th>Case</th>
<th>Sex</th>
<th>Family history</th>
<th>Onset (yrs.)</th>
<th>Death (yrs.)</th>
<th>Duration (yrs.)</th>
<th>Histopathological type</th>
</tr>
</thead>
<tbody>
<tr>
<td>1. K.H.</td>
<td>M</td>
<td>Noncontributory</td>
<td>15</td>
<td>17</td>
<td>2</td>
<td>Lafora-body type</td>
</tr>
<tr>
<td>2. C.M.</td>
<td>F</td>
<td>Noncontributory</td>
<td>13</td>
<td>17</td>
<td>4.7</td>
<td></td>
</tr>
<tr>
<td>4. M.T.</td>
<td>F</td>
<td>Noncontributory</td>
<td>2</td>
<td>16</td>
<td>14</td>
<td></td>
</tr>
<tr>
<td>5. T.T.</td>
<td>M</td>
<td>Sibling</td>
<td>18</td>
<td>39</td>
<td>21</td>
<td></td>
</tr>
<tr>
<td>6. S.T.</td>
<td>M</td>
<td>Noncontributory</td>
<td>15</td>
<td>26</td>
<td>11</td>
<td></td>
</tr>
<tr>
<td>7. S.N.</td>
<td>M</td>
<td>Noncontributory</td>
<td>7</td>
<td>38</td>
<td>31</td>
<td></td>
</tr>
<tr>
<td>8. T.N.</td>
<td>F</td>
<td>Noncontributory</td>
<td>13</td>
<td>41</td>
<td>28</td>
<td></td>
</tr>
<tr>
<td>9. K.I.</td>
<td>M</td>
<td>Noncontributory</td>
<td>13</td>
<td>24</td>
<td>11.2</td>
<td>Lipidosis type</td>
</tr>
<tr>
<td>10. M.Y.</td>
<td>M</td>
<td>Sibling</td>
<td>17</td>
<td>22</td>
<td>5</td>
<td></td>
</tr>
<tr>
<td>11. T.Y.</td>
<td>M</td>
<td>Similar disease in sibling</td>
<td>13</td>
<td>18</td>
<td>5</td>
<td></td>
</tr>
<tr>
<td>12. H.E.</td>
<td>M</td>
<td>Similar disease in sibling</td>
<td>11</td>
<td>21</td>
<td>10</td>
<td></td>
</tr>
</tbody>
</table>

Special type
cells (case 1, 3, 4) also the similar degenerated substance were demonstrated, but not found at all in case 2. And then, case 3 could be diagnosed at liver biopsy (Inoue et al., 1974) and intranuclear inclusions of astrocytes of Alzheimer’s II type, which were not related to Lafora-body type, could be noted in case 4 with hepatic damage-fatty liver (Kotorii et al., 1976).

Then the basophilic inclusions reported by Lafora et al. (1911) were considered to be the secondary products as a result of histochemical investigation and electron microscopical examination. The primary products of the Lafora-bodies were not basophilic but positive with Best’s carmine stain and

Lafora-body type

*Fig. 1* case 3: Lafora-bodies in the dentate nucleus. Nissl’s stain.

*Fig. 2* case 3: PAS-Positive substances in the liver cells (liver biopsy).

*Fig. 3* case 4: Lafora-body (↑) and intranuclear inclusions (glycogen) of astrocytes (↑↑). Best’s carmine stain.

*Fig. 4* case 4: Fatty liver. H. E. stain.
electron microscopically were composed of fibrils which were about 100 Å in diameter. Histochemical investigation on the Lafora-bodies showed to contain mucopolysaccharides or acid mucopolysaccharides (Anraku et al., 1966; Kotorii et al., 1974). But the mechanism of formation of the Lafora-body is still unknown.

2. Degenerative type (case 5-case 8) : The histopathological changes, which consisted of non-specific degenerative changes, i.e. neuronal loss, simple neuronal atrophy and gliosis, were most marked in the dentate nucleus and the cerebellar cortex, with less marked changes in the olivary nucleus. However, it should be considered that the lesion of the cerebellar cortex may be relevant to repeated convulsions and long-term administration of diphenylhydantoin. The cerebral cortex, Ammon's horn and the nucleus cerebri etc., were scarcely involved.

Lafora-body was not found at all in the central nervous system.

3. Lipidosis type - Gaucher's disease (case 9) : Main pathological findings were splenomegaly and enlarged mesenteric lymph nodes in both of which Gaucher's cells containing histochemically demonstrable glycolipids were observed to be present.

At autopsy findings in the brain, macroscopically there were no conspicuous alterations. Microscopically neuronal loss and neuronophagia in the dentate nucleus were noted. A histo-

Special type

Fig. 5 case 10 : Pigments (green - dark green) in the upper layer of the cerebral cortex. Nissl's stain.

Fig. 6 case 10 : Pigments (green - dark green) in the layer of Purkinje cells. Nissl's stain.
SHORT COMMUNICATION

A pathological finding of particular note was vascular changes characterized by conspicuous enlargement and proliferation of adventitial cells. Some of these cells bore resemblance to Gaucher's cell and presented histochemical findings suggestive of the presence of glycolipids. Moreover, proliferation of blood vessels was prominent in the amygdaloid nucleus (Anraku et al., 1973).

4. Special type (case 10 - case 12): Non-specific degenerative changes of nerve cells were more insignificant than those of degenerative type.

As the most striking changes common to all these 3 cases observed, however, a large number of various brown granular pigments could be seen lying almost free in the specific areas involving the upper layer of the cerebral cortex, the layer of purkinje cells, the globus pallidus and the substantia nigra.

Histochemical investigation on these pigments revealed that there existed argyrophil substances combined with protein, but the presence of lipids could not be confirmed. These findings were different from a special form of amaurotic idiocy reported by Moschel (1954) and Seitelberger (1962) (Anraku et al., 1965).

REFERENCES


