Dentatorubral-pallidoluysian Atrophy (DRPLA; Haw River Syndrome; Natito-Oyanagi disease) – OMIM 125370

**Background**

DRPLA is a progressive brain disorder. Onset can vary – juvenile-onset (<20 years old), early adult-onset (20-40 years old) and late adult-onset (>40 years old) – although the average age for onset is around 30. Symptoms vary depending upon the age of onset; children display seizures, involuntary muscle twitching and intellectual disability, whereas in adults the symptoms include dementia, ataxia and psychiatric problems. DRPLA is caused by a mutation in the gene *ATN1* (OMIM 607462), and follows an autosomal dominant inheritance pattern. Despite a number of sporadic cases in Western countries, this disease appears to be rare except in Japan.

**Recommended Clinical Referral Criteria**

- Diagnostic cases and at risk family members
- Autosomal dominant inheritance
  - Onset <20 years with ataxia, myoclonus, seizures and progressive intellectual deterioration
  - Onset >20 years with ataxia, choreoathetosis, dementia and psychiatric disturbance

**Molecular Analysis**

<table>
<thead>
<tr>
<th>Molecular Strategy</th>
<th>Mutation Detected</th>
<th>Test Clinical Sensitivity</th>
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</thead>
<tbody>
<tr>
<td>Fragment length analysis of <em>ATN1</em> gene expansion</td>
<td><em>ATN1</em> gene [CAG]n expansion</td>
<td>100% for patients meeting clinical diagnostic criteria</td>
</tr>
</tbody>
</table>

**Prices & Turnaround Times (TAT)**

This test is carried out by the molecular laboratory in Cambridge – please click on the link below for further details:

[http://www.cuh.org.uk/addenbrookes/services/clinical/genetics/labs/services_test/molecular_genetics_tests.html](http://www.cuh.org.uk/addenbrookes/services/clinical/genetics/labs/services_test/molecular_genetics_tests.html)