ANDERSEN-TAWIL SYNDROME (ATS) KCNJ2 (OMIM: 600681)

Introduction
Andersen-Tawil syndrome (ATS) is a multisystem channelopathy characterised by a triad of periodic paralysis, ventricular arrhythmias, and distinctive dysmorphic facial or skeletal features. ATS is inherited in an autosomal dominant pattern with intra-familial variable expression and incomplete penetrance. It is predisposed to by mutations in the KCNJ2 gene\[a-d]\.

REFERRAL PROCEDURE
• Diagnostic referrals are accepted for probands with a suspected or confirmed diagnosis of Brugada syndrome.
  o Referrals are accepted from Cardiology, Clinical Genetics and other relevant medical specialities.
  o Clinical information and details of relevant family history should be provided with all referrals either on the original request form or on a separate pre-referral form (Cardiac arrhythmia pre-referral form).
• Family test referrals are only accepted from Clinical Genetics specialists.
  o Referrals for affected family members (i.e. segregation analysis) must be accompanied by appropriate clinical information.
  o Referrals for unaffected family members will only be considered for variants with clear evidence for pathogenicity.
• Clinical advice is available from Dr Edward Blair, Consultant Clinical Geneticist, at the Churchill Hospital (Ed.Blair@ouh.nhs.uk).
• Further information about the test can be obtained from the laboratory (OxfordCardiac@nhs.net)

Sample Requirements
• This analysis requires 0.5 µg of DNA, preferably at >100 ng / µl.
• DNA from blood or post-mortem tissues is suitable.

Testing Strategy
Molecular analysis is undertaken by fluorescent dye-deoxy sequencing of the coding exon and intron/exon boundaries of KCNJ2.

Target Reporting Times

<table>
<thead>
<tr>
<th>Referral Type</th>
<th>Molecular Analysis</th>
<th>Target Reporting Time (working days)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Diagnostic</td>
<td>Sequence analysis of KCNJ2</td>
<td>40</td>
</tr>
<tr>
<td>Familial</td>
<td>Familial mutation(s)</td>
<td>10</td>
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</tbody>
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• Please see the current price list on the Oxford Genetics laboratory Molecular genetics page


N.B. Details are correct for the date of printing only. Last updated August, 2015