Identifying Patients

**WHO MAY BENEFIT FROM GENETIC TESTING**

**Practice Guideline**

American College of Medical Genetics and Genomics (ACMG) | American Academy of Pediatrics (AAP) | American Academy of Neurology (AAN)

The ACMG, AAP, and AAN recommend chromosomal microarray (CMA) as a first-tier genetic test in the postnatal evaluation of individuals with ID and/or ASD.

For patients with ID and/or ASD, fragile X syndrome testing may be considered along with CMA.

Adapted from Miller DT, et al., *Am J Hum Genet*, 2010

Ambry offers a range of first-tier tests and multi-gene panels for patients with neurodevelopmental disorders. If your patient has any of the following disorders, these genetic tests may be considered.

<table>
<thead>
<tr>
<th>DISORDER</th>
<th>FIRST-TIER</th>
<th>TARGETED PANEL</th>
<th>COMPREHENSIVE</th>
</tr>
</thead>
<tbody>
<tr>
<td>INTELLECTUAL DISABILITY (ID)</td>
<td>Chromosomal microarray and fragile X testing</td>
<td>IDNext</td>
<td>ExomeNext</td>
</tr>
<tr>
<td>AUTISM SPECTRUM DISORDERS (ASD)</td>
<td>Chromosomal microarray and fragile X testing</td>
<td>AutismNext</td>
<td>ExomeNext</td>
</tr>
<tr>
<td>ID/ASD IN CONJUNCTION WITH OR WITHOUT EPILEPSY</td>
<td>Chromosomal microarray</td>
<td>Neurodevelopment-Expanded</td>
<td>ExomeNext</td>
</tr>
</tbody>
</table>

We know your time is valuable. Now you can reduce follow-up by submitting parental samples (biological mother and father) along with the patient sample. Co-segregation studies will be performed as needed prior to results being released.
Choosing the Right Test

**Based on Clinical Presentation**

- **IDNext**
  - 140 Genes
  - TAT: 2-3 weeks

- **EpilepsyNext**
  - 100 Genes
  - TAT: 2-3 weeks

- **AutismNext**
  - 48 Genes
  - TAT: 2-3 weeks

Neurodevelopment-Expanded includes all 196 genes in these three tests.

**TAT:** 4-6 weeks.
Just as no two fingerprints are alike, the way disease presents itself in every individual is different. Since 1999, our mission has always been about understanding disease better, so treatments and cures can be found faster. Every sample that arrives in our lab is viewed as a person with a life and a story that is unique to only them. By providing advanced confirmation genetic testing for inherited and non-inherited diseases, we can help you make more informed and responsible treatment decisions with your patients.