Ambry Genetics®
A Konica Minolta Company

Neurodevelopmental Disorders Genetic Testing

REFERENCE GUIDE

February 2019

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.

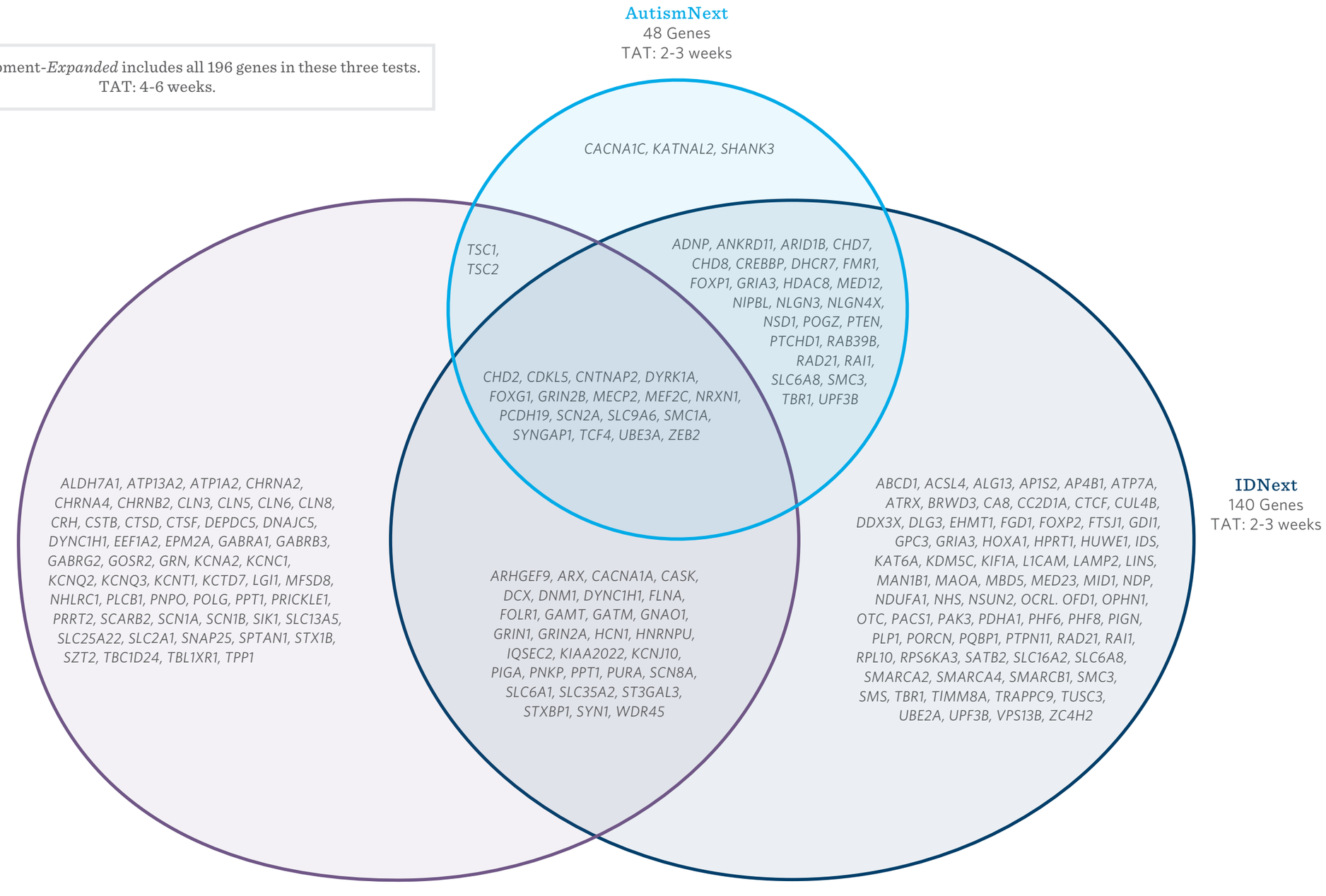
Adapted from Miller DT, *et al.*, Am J Hum Genet., 2010

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.

DISORDER	FIRST-TIER	TARGETED PANEL	COMPREHENSIVE
INTELLECTUAL DISABILITY (ID)	Chromosomal microarray and fragile X testing	IDNext	ExomeNext
AUTISM SPECTRUM DISORDERS (ASD)	Chromosomal microarray and fragile X testing	AutismNext	ExomeNext
ID/ASD IN CONJUNCTION WITH OR WITHOUT EPILEPSY	Chromosomal microarray	Neurodevelopment-*Expanded*	ExomeNext

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.

Neurodevelopment-*Expanded* includes all 196 genes in these three tests.
TAT: 4-6 weeks.



Over 1 Million Tests Completed

MOVING SCIENCE FORWARD

Purposeful Confirmatory Testing

Many labs validate their tests based on certain limited studies. That's why we participated and led the largest study of its kind (20,000 cases) guiding us to utilize confirmatory testing when we see specific well-defined thresholds. Our mission is to get it right the first time.

Understanding Disease Better Through Free Data Sharing

Identifying an individual's genetic information is nothing new—it's what we do with it that is unique. When labs share genomic information, we can together accelerate the understanding of human disease. Through AmbryShare, we leverage de-identified information to collaborate with others and help people everywhere find answers.

SuperLab

Our 65,000 square foot highly-automated CLIA/CAP certified lab produces some of the fastest turnaround times in the industry, without compromising testing accuracy or specificity.

Ambry's Translational Genomics (ATG) Lab

As an advanced diagnostic lab, it's our responsibility to ensure the results you get from us are accurate and that classification is as complete and robust as possible. Our ATG lab is a unique laboratory that provides an additional service at no additional cost for you and your patients to generate more precise data potentially bringing clarity to some variants of unknown significance (VUS). This helps to actively drive down the rate of VUS results and can give you an increased understanding of your patient's results, so you can better provide medical management recommendations and improve health outcomes.

About Ambry

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.

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