



EPIDASD

Epilepsy – Intellectual Disability – Autism Spectrum Disorder

Highlights

- The screening panel designed by Amplexa Genetics consists of 1740 different genes related to these neurological diseases.
- Rare or low-frequency variants are evaluated according to ACMG and ACGS guidelines.
- EPIDASD helps your patients to have an early diagnosis facilitating a personalized treatment. [1]
- The genes included in the EPIDASD panel are selected based on clinical evidence, scientific publications, the Human Gene Mutation Database (HGMD) and the Online Mendelian Inheritance in Men (OMIM).



EPIDASD

Epilepsy – Intellectual Disability – Autism Spectrum Disorder

We support you

- With fast turn-around times.
- With high quality clinical reports with easy to read summary of results.
- With easy access to skilled doctors and molecular geneticists helping you to interpret the findings.

First step towards precision medicine is precision diagnosis!



Amplexa Genetics®



NEUROLOGY

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Amplexa Genetics®

Providing knowledge

EPIDASD

Epilepsy, Intellectual Disability and Autism Spectrum Disorders

The most common neurological and neuropsychological diseases.

INTRODUCTION

The EPIDASD panel consists of 1740 genes known to be involved in the development of epilepsy, developmental and epileptic encephalopathies, intellectual disability syndromes, brain malformations and/or autism spectrum disorders.

The test is relevant for patients suffering from epilepsy, intellectual disability and/or autism spectrum disorder with or without epilepsy.

Although any patient with epilepsy may benefit from genetic testing, screening with the EPIDASD panel will be of greatest importance to patients with syndromic epilepsy and patients with early-onset seizures (less than 3 years of age), and/or a family history of seizures, neurological deficit, an autism spectrum disorder, or intellectual disability. [2]

THE ANALYSIS

In total, 1740 genes associated with Epilepsy, Intellectual Disability, and Autism Spectrum Disorders are analyzed in the EPIDASD panel. Data is extracted as a virtual panel from human exome data, and an in-house bioinformatic pipeline performs variant calling and filtering.

Rare or low-frequency variants (SNVs and CNVs) are evaluated according to the ACMG guidelines [3], the ACGS guidelines as well as individual estimates.

In the evaluation are database searches in the Human Gene Mutation Database (HGMD), ClinVar, DECIPHER, the Genome Aggregation Database (gnomAD, release 4.0.0), and dbSNP (155).

Synonymous variants in autosomal dominant genes which have been observed more than 3 times or in homo-/hemizygous state in the gnomAD database are not reported.

Predictions on protein level are obtained from dbNSFP Functional Predictions and Cores 3.0 database, REVEL functional prediction, and CADD score 1.6.

Predictions on transcriptional level variants are submitted to bioinformatics software tools e.g. NNSplice, GeneSplicer, MaxEntScan and PWN.

General Specifications

Analysis method:	Next Generation Sequencing
Hardware:	Illumina NovaSeq 6000
Chemistry:	Twist Human Core Exome
Sample Type:	DNA, saliva, blood, tissue
Nomenclature:	According to HGVS
Reference database:	RefSeq

Scan the EPIDASD genes here:



EPIDASD GENE LIST:
1740 GENES

EVALUATION

Reported variants

Class III	Uncertain Significance
Class IV	Likely Pathogenic
Class V	Pathogenic

EPIDASD

Average read depth	70-100x
Target sequence covered >20x	>97%

LIMITATIONS OF THE ANALYSIS

The analysis has been limited to protein coding regions and 10bp from the exon-intron boundaries including splice sites. Non-coding regions: 5'-UTR, 3' UTR, introns, and promoter regions have been sparsely analyzed, and large rearrangements will not be detected.

The method is also not appropriate for the analysis of repeat regions. For analysis of the FMR1 gene we recommend our FMR1- Fragile X (CGG) repeat expansion test. Only associations with phenotypes relevant to the indication for choosing the selected panel are reported.

REFERENCES

- [1] Brunklaus et al., (2020), Biological concepts in human sodium channel epilepsies and their relevance in clinical practice PMID: 32090326.
- [2] Bayat et al., (2022), Impact of Genetic Testing on Therapeutic Decision-Making in Childhood-Onset Epilepsies—a Study in a Tertiary Epilepsy Center, PMID: 35723786.
- [3] Richards et al., (2015), Standards and guidelines for the interpretation of sequence variants: a joint consensus recommendation of the American College of Medical Genetics and Genomics and the Association for Molecular Pathology, PMID: 25741868

SUMMARY AMPLEXA NEUROLOGY

- Coding Sequences
- Mitochondrial genome
- >100x highly uniform coverage
- >10 years of experience
- >40 scientific publications
- External Quality Assurance schemes for all our methods

