Invitae's mission is to bring comprehensive genetic information into mainstream medicine to improve healthcare for billions of people.

Driving down the cost of genetic information will increase its personal and clinical utility

Healthcare professionals are fundamental in ordering and interpreting genetic information

People should own and control their own genetic information

Genetic information is more valuable when shared

Enhancing the Experience for Referring Clinicians

Board-certified genetic counselors, laboratory geneticists, and clinical geneticists are available to discuss testing needs, test results, and research collaborations.

Flexible ordering - Clinicians can order single genes, small panels, or large panels in any combination suited to patients' diagnostic needs.

Re-requisition - After receiving a negative result, clinicians can request results from additional genes related to the presenting phenotype within 90 days.

Family variant testing - Requested variant testing of likely pathogenic and pathogenic variants available for all blood relatives of a proband with panel results obtained from Invitae.

Adult Epilepsy

Received: 12 December 2018 | Revised: 28 May 2019 | Accepted: 7 June 2019
DOI: 10.1111/epi.16273

FULL-LENGTH ORIGINAL RESEARCH

Epilepsia

Clinical utility of multigene panel testing in adults with epilepsy and intellectual disability

Felippe Borlot[1,2] | Bruno Ivo de Almeida[1,3] | Shari L. Combe[1] | Danielle M. Andrade[2,4,5] | Francis M. Filloux[1] | Kenneth A. Myers[6,7]

Received: 11 March 2019 | Revised: 19 May 2019 | Accepted: 31 May 2019
DOI: 10.1002/epi4.12348

FULL-LENGTH ORIGINAL RESEARCH

Epilepsia Open® Open Access

Possible precision medicine implications from genetic testing using combined detection of sequence and intragenic copy number variants in a large cohort with childhood epilepsy

Rebecca Truty[1] | Nila Patil[2] | Raman Sankar[2] | Joseph Sullivan[3] | John Millichap[4] | Gemma Carvill[5] | Ali Entezam[1] | Edward D. Esplin[1] | Amy Fuller[1] | Michelle Hogue[1] | Britt Johnson[1] | Amirah Khouzam[1] | Yuya Kobayashi[1] | Rachel Lewis[1] | Keith Nykamp[1] | Darlene Riethmaier[1] | Jody Westbrook[1] | Michelle Zeman[1] | Robert L. Nussbaum[1,6] | Swaroop Aradhya[1]

1. Adults with epilepsy and intellectual disability

- PMID 31273778: Epilepsy panel testing identified an etiology in 22% of adults with epilepsy and intellectual disability.

2. Adults with poorly controlled seizures

- PMID 31440721: Precision medicine implications (PMIs) were immediately relevant to >33% of individuals with a positive molecular diagnosis (PosMD). Of those with PosMDs specifically related to PMIs, 66% were in children younger than 5 years, 27% in children aged 5-17 years, and the remaining 7% in adults.

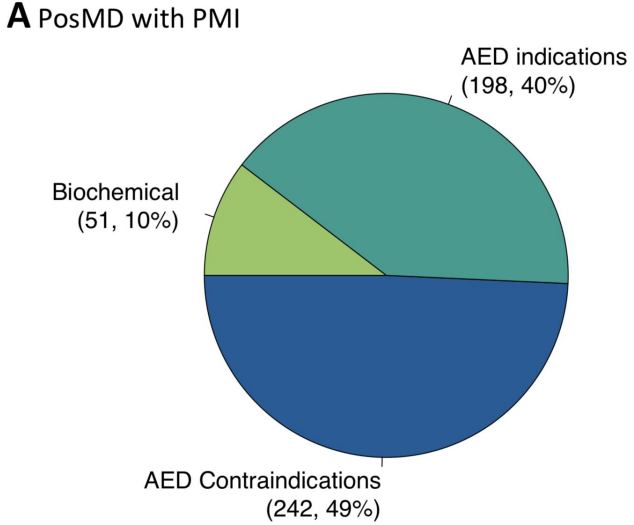
3. Adults who are seeking out information regarding recurrence risk -- informed about their chances to pass down a susceptibility to seizures/epilepsy to their children

4. Adults with epilepsy "plus" clinical presentation (e.g., epilepsy + intellectual disability, autism spectrum disorder, syndromic presentation, etc.)

INVITAE

Categories of Precision Medicine Implications

- Nearly **half of positive molecular diagnoses with precision medicine implications were related to contraindications** for antiepileptic drugs, largely due to variants in SCN1A.
- **10% of molecular diagnoses were related to biochemical disorders** with available treatments.
- 40% of molecular diagnoses invoked indications for specific anti-epileptic drugs (e.g. Vigabatrin for spasms in TSC).
- Another **21% of individuals** had positive molecular diagnoses in **genes with emerging association** with precision medicine implications.



Family Variant Testing

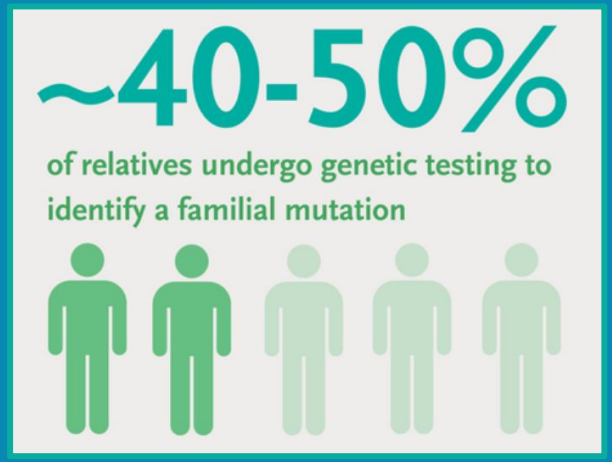
When a patient tests positive through one of Invitae's diagnostic panel tests, family variant testing is available at no additional charge to eligible family members.

FVT policy temporarily extended from 90 days to 150 days

An example of our commitment to lowering barriers to genetic testing.

Genetic insights are a family affair

- Identifying the presence of a variant can sometimes enable medical management
- Identifying the absence of a variant can also have significant implications for relatives.



INVITAE

https://www.invitae.com/en/prepared/

Leveraging Telehealth Capabilities

In the clinic or at home, we're here to help.

Our genetic testing services are supported with telehealth options.

Most Invitae genetic tests can be completed with a saliva sample kit shipped directly to your patient.

Our family variant testing and re-requisition policies have been extended to 150 days from 90 days during this time.

Perspectives in Genetics

"Seeing patients from home (and staying HIPAA-compliant): A guide for GCs"

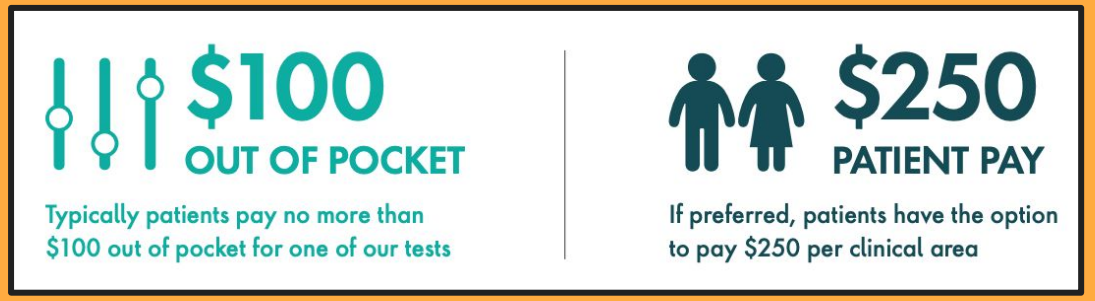
No Surprises. Simple Billing.

Invitae is proud to be in-network for more than 300 million patients in the US - and growing.

We also offer a generous patient assistance program - click **here**.

Only approximately 1% of genetic tests received at Invitae are cancelled due to financial concerns.

Transparent pricing, ethical billing



*After billing insurance, the majority of patients pay $100 or less, but that amount could be higher or lower depending on the plan.

Want more details about billing at Invitae? Click here.