XOMEDXXPRESS AND XOMEDXPRIORITY

RAPID EXOME TESTING

XomeDxXpress and XomeDxPriority are expedited clinical exome sequencing services provided by GeneDx.

OPTIONS FOR EXPEDITED CLINICAL EXOME SEQUENCE TESTING

XomeDxXpress and XomeDxPriority

- Trios (proband and both biological parents) are preferred; non-trios must be approved by GeneDx
- Fresh blood samples are the preferred specimen type
- Institutional or Self-Pay only
- Client must inform GeneDx of the case prior to samples arriving
- Use Xpress@GeneDx.com for both XomeDxXpress and XomeDxPriority testing

XomeDxXpress

- Within 7 calender days after the start of testing, receive a verbal result of pathogenic and/or likely pathogenic variants in known disease-causing genes (Human Genome Mutation Database genes)
- 14 day turnaround time for a written report for all confirmed variants

XomeDxPriority

- Once test is activated in the lab, it is a 4 week turnaround time for a written report for all confirmed variants

WHEN IS XOMEDXXPRESS OR XOMEDXPRIORITY USEFUL?

Useful for any patient for whom a rapid molecular diagnosis may direct or alter medical management. Appropriate scenarios include an affected individual with:

- Rapidly deteriorating clinical status
- Multiple congenital anomalies
- High-acuity illness manifesting with seizures, hypotonia and morphological abnormalities of the central nervous system
- A genetic syndrome or underlying metabolic disorder
- A clinical presentation for which a molecular diagnosis may eliminate the need for further invasive testing
- A disease which is highly genetically heterogeneous and no single gene or groups of genes makes up a significant percentage of the mutation spectrum

REQUIREMENTS FOR XOMEDXXPRESS AND XOMEDXPRIORITY:

- Ordering providers must contact GeneDx prior to sending the samples by emailing: Xpress@genedx.com
- **Specimen:** Blood is the preferred specimen type. If blood is unavailable, the provider must discuss this with the Xpress team.
- We prefer to receive trios (samples on the proband and both biological parents). If a trio is not available, the provider must discuss this with the Xpress team.
- We prefer to receive a copy of the requisition, signed consent forms and relevant clinical information prior to receiving the samples.
- **Billing Options:** Institutional or Self-Pay

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CASE REPORT:

A young male presents with developmental delay, hypotonia, failure to thrive, respiratory failure, hepatosplenomegaly, and dolichocephaly. No previous genetic testing was performed.

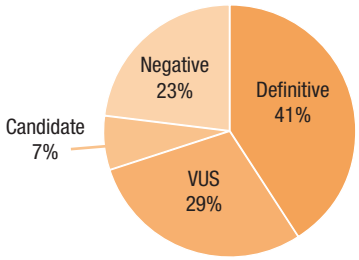
XomeDxXpress testing revealed a dual diagnosis in this patient, compound heteroxygous variants in the NPC1 gene (Neimann-Pick C) and a de novo variant in the KMT2A gene (Wiedemann-Steiner syndrome).

Verbal results were provided in 5 days and the written reported completed in 10 days.

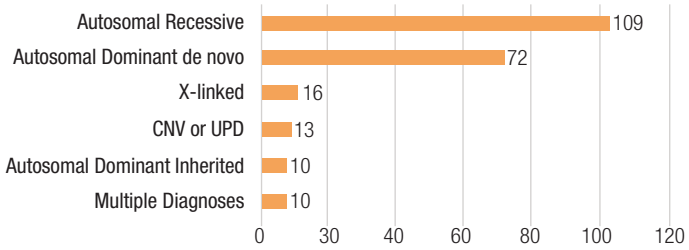
The family can consider new drug therapies and enrollment in the Cyclohexadiene trial. Targeted testing can be offered to asymptomiatic siblings and prenatal diagnosis is now an option for future pregnancies.

XOMEDXXPRESS TESTING OUTCOMES

Diagnostics Yield of XomeDxXpress (n=500)

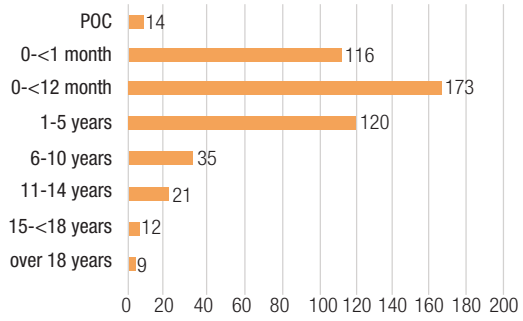


Modes of Inheritance in Positive Cases

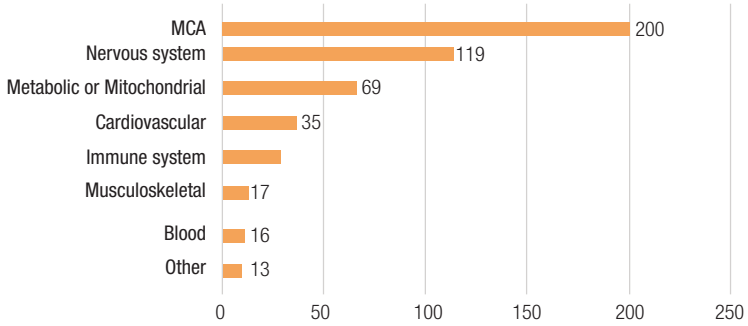


PATIENT POPULATION

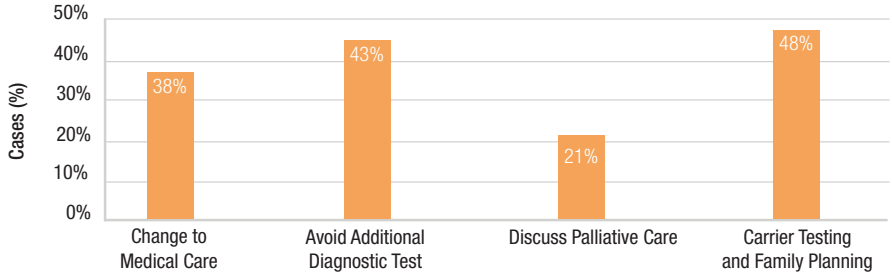
Proband Age during XomeDxXpress



Primary Indications for XomeDxXpress Testing



Provider utilization of results from 58 XomeDxXpress case



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