XOMEDxXPRESS AND XOMEDxPRIORITY
RAPID EXOME TESTING

XomeDxXpress and XomeDxPriority are clinical exome sequencing services provided with a quicker turnaround time when timing is critical to possibly direct or alter medical management.

WHEN IS XOMEDxXPRESS OR XOMEDxPRIORITY USEFUL?

May be useful in patients with any of the following:

- 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

OPTIONS FOR EXPEDITED CLINICAL EXOME SEQUENCE TESTING

XomeDxXpress

Verbal result of pathogenic and/or likely pathogenic variants in known disease causing genes (Human Genome Mutation Database genes) within **7 calendar days** after the start of testing

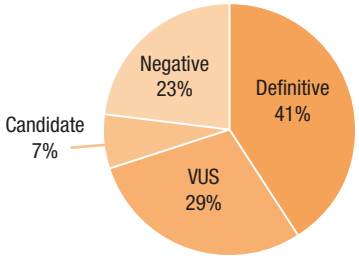
Written report for all confirmed variants within **14 days** after start of testing

XomeDxPriority

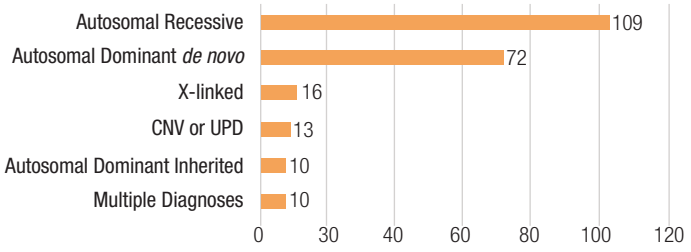
Written report for all confirmed variants within **4 weeks** after start of testing

XOMEDxXPRESS TESTING OUTCOMES

Diagnostic Yield of XomeDxXpress (n=500)



Modes of Inheritance in Positive Cases



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CASE REPORT:

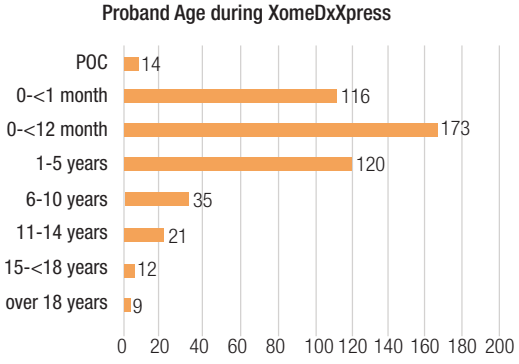
A 34-month old male was referred with developmental delay, hypotonia, failure to thrive, respiratory failure, hepatosplenomegaly, and dolichocephaly. No previous genetic testing was performed. Patient had been hospitalized in the PICU for months with a tracheostomy and G-tube. A muscle biopsy and many other tests were previously performed to try to make a diagnosis.

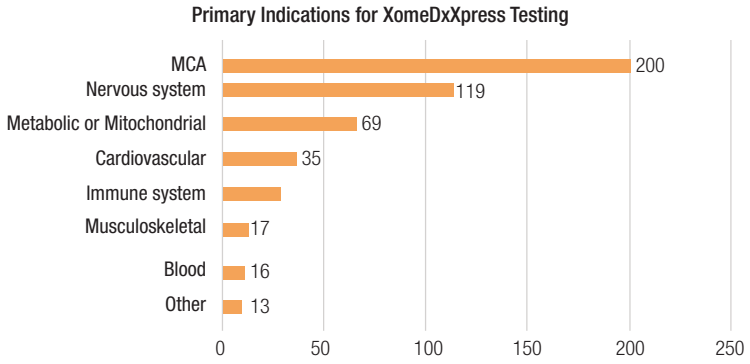
XomeDxXpress testing revealed a dual diagnosis in this patient, compound heteroxygous variants in the *NPC1* gene (Neimann-Pick C) and a *de novo* variant in the *KMT2A* gene (Wiedemann-Steiner syndrome).

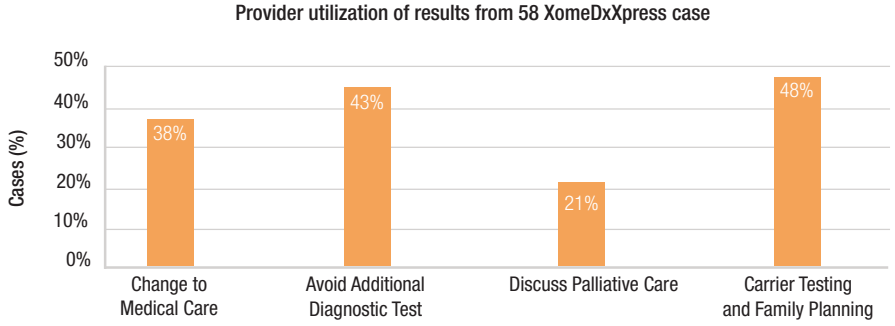
Verbal results were provided in 5 days and the written reported completed in 10 days.

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

PATIENT POPULATION







REQUIREMENTS FOR XOMEDxXPRESS AND XOMEDxPRIORITY:

If any of these are not available/possible, please discuss with the Xpress team.

- Ordering providers must contact GeneDx prior to sending the samples by emailing: Xpress@genedx.com
- Fresh blood samples are the preferred specimen type.
- We prefer to receive trios (samples on the proband and both biological parents).
- Please send a copy of the requisition, signed consent forms and relevant clinical information prior to sending the samples, if possible.
- Institutional or Self-Pay only.

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