Rare Disease Genetic Testing Support Programs

One of the key challenges associated with launching therapeutics in the rare and orphan disease space is identifying patients that have the conditions that these therapies can address. This includes issues with motivating physicians to order tests and labs to offer testing.

One solution has been the deployment of sponsored testing support programs, which facilitate broad access by alleviating the need to secure payer reimbursement for testing.

Examples of Testing Support Programs

Sanofi Genzyme *Lumizyme for Pompe Disease*	*Sanofi Genzyme finances a collaboration between the Muscular Dystrophy Association and Emory Genetics Laboratory*	Free testing is available for the 30 different genetic subtypes for patients with unexplained limb-girdle muscle weakness which may be caused by Pompe disease
CSL Behring *Zemaira for Alpha-1 Antitrypsin Deficiency (AATD)*	*CSL Behring funds a DNA1 Alpha-1 testing program using a next-generation sequencing (NGS) test by Biocerna*	DNA1 screens for alpha-1 antitrypsin by sequencing the SERPINA1 gene to help identify known and unknown clinically relevant variants causing AATD
Alnylam *Patisiran for Hereditary Transthyretin Amyloidosis (hATTR)*	*Alnylam launched the Alnylam Act program in partnership with Invitae to offer free genetic testing and counseling*	Free genetic testing is available for patients suspected with hATTR, as well as those with confirmed family history of hATTR to aid in patient identification
Alexion *Rare Genetic Disorders*	*Alexion partnered with Rady's Children's Institute for Genomic Medicine to develop a SmartPanel with Sema4*	SmartPanel leverages patient presentation information and Alexion's rare disease database to create an open platform to provide automated rare disease diagnoses

The Bottom Line

For indications where genetic testing can unlock access to therapies, **support programs may serve a key role in enhancing diagnosis rates**.

As the use of these programs expands, **future bundling of targeted genetic testing or newborn screening via advanced tools** may move these programs beyond targeted therapeutic areas. This could further enhance potential to **improve patient identification despite potential access barriers**.

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