

## Carrier Screening Super Panel

Expanded screening to improve your patient outcomes

### The SDxLabs Super Panel

At SDxLabs, part of our mission is to improve pregnancy and newborn outcomes. We achieve this by offering products that follow evidence-based societal guidelines such as those provided by the American College of Medical Genetics (ACMG) to provide actionable results. The Super Panel carrier screen focuses on giving your patients equitable care regardless of their race or ancestry by using a specifically curated gene list and full gene sequencing technology.

145

Conditions

The Super Panel screens for 145 conditions that are relevant and actionable for all ethnicities

1 in 23

At-Risk

The Super Panel's at-risk couple detection rate is 1 in 23

## A case study in expanded carrier screening

### Medium-Chain Acyl-CoA Dehydrogenase Deficiency (MCADD)

#### Early intervention prevents mortality and morbidity

- 1 in 66 people are a carrier of MCADD. If both parents are carriers, there is a 25% chance a child will be affected by this disorder.
- Babies who have MCADD are unable to break down medium-chain fatty acids, which can lead to metabolic decomposition.
- 5-9% of babies with MCADD will pass away while waiting for newborn screening results.<sup>3-7</sup>
- Treatment for this condition is as simple as feeding the newborn every four hours.
- Screening for MCADD with the Super Panel allows affected babies to receive treatment right from birth.



1 in 300 pregnancies are impacted by recessive and X-linked conditions<sup>1</sup>



Genetic conditions become more treatable or manageable when couples know early



80% of babies born with a genetic condition have no known family history of it<sup>2</sup>



# Why choose SDxLabs?

**100%**  
of Patients

## Carrier Screening is for all patients

- Autosomal recessive and X-linked conditions impact 1 in 300 pregnancies<sup>1</sup>
- Early identification of at-risk couples allows for early interventions and reproductive planning

**>99%**  
Detection  
Rate

## Not all approaches to carrier screening provide equitable care

- Screening for CF and SMA alone or using ethnicity-based screening misses carriers for other conditions
- The Super Panel closely aligns with the ACMG guidelines for ethnicity-neutral screening and has a >99% detection rate for most conditions<sup>8</sup>

**145**  
Conditions

## Federally recommended conditions

- The Super Panel screens for 145 genetic conditions
- 35 of these conditions are federally recommended for newborn screening, minimizing surprises at birth

**2**  
Weeks

## Fast turnaround time

- Results in approximately 2 weeks from sample receipt

## Support and Service

SDxLabs' board-certified genetic counselors help you and your patients understand carrier screening and the possible results it can give. Post screen, they can review results with your patient and help plan next steps. Patients can reach out to our Customer Care team for questions related to billing, insurance coverage, or the status of results.

### Genetic Counseling

(412) 677-0664 Option 3  
Genetics@SignatureDx.com

### Customer Care

(412) 677-0664 Option 1  
CustomerCare@SignatureDx.com



## About SDxLabs

SDxLabs delivers noninvasive diagnostic and women's health solutions that support proactive healthcare decisions. Our services focus on early detection and actionable results that enable effective treatments and improved patient outcomes. SDxLabs is committed to empowering patients, providers, and the community with access to relevant health services and unmatched service.

1501 Preble Avenue, Suite 200 Pittsburgh, PA 15233 | (412) 677-0664 | [SDx-Labs.com](https://www.sdx-labs.com)

## References

1. Johansen Taber, K. A., Beauchamp, K. A., Lazarin, G. A., Muzzey, D., Arjunan, A., & Goldberg, J. D. (2019). Clinical utility of expanded carrier screening: Results-guided actionability and outcomes. *Genetics in Medicine*, 21(5), 1041-1048. <https://doi.org/10.1038/s41436-018-0321-0>
2. Blythe, S. A., & Farrell, P. M. (1984). Advances in the diagnosis and management of cystic fibrosis. *Clinical Biochemistry*, 17(5), 277-283. [https://doi.org/10.1016/S0009-9120\(84\)90541-1](https://doi.org/10.1016/S0009-9120(84)90541-1)
3. Ahrens-Nicklas, R. C., Pyle, L. C., & Ficioglu, C. (2016). Morbidity and mortality among exclusively breastfed neonates with medium-chain acyl-CoA dehydrogenase deficiency. *Genetics in Medicine*, 18(12), 1315-1319. <https://doi.org/10.1038/gim.2016.49>
4. Ensenauer, R., Winters, J. L., Parton, P. A., Kronn, D. F., Kim, J., Matern, D., Rinaldo, P., & Hahn, S. H. (2005). Genotypic differences of MCAD deficiency in the asian population: Novel genotype and clinical symptoms preceding newborn screening notification. *Genetics in Medicine*, 7(5), 339-343. <https://doi.org/10.1097/01.GIM.0000164548.54482.9D>
5. Wilcken, B., Prof, Haas, M., PhD, Joy, P., PhD, Wiley, V., PhD, Chaplin, M., BAppSc, Black, C., BPsych, Fletcher, J., MD, McGill, J., MBBS, & Boneh, A., MD. (2007). Outcome of neonatal screening for medium-chain acyl-CoA dehydrogenase deficiency in australia: A cohort study. *The Lancet (British Edition)*, 369(9555), 37-42. [https://doi.org/10.1016/S0140-6736\(07\)60029-4](https://doi.org/10.1016/S0140-6736(07)60029-4)
6. Lovera, C., Porta, F., Caciotti, A., Catarzi, S., Cassanello, M., Caruso, U., Gallina, M. R., Morrone, A., & Spada, M. (2012). Sudden unexpected infant death (SUDI) in a newborn due to medium chain acyl CoA dehydrogenase (MCAD) deficiency with an unusual severe genotype. *Italian Journal of Pediatrics*, 38(1), 59-59. <https://doi.org/10.1186/1824-7288-38-59>
7. Andresen, B. S., Lund, A. M., Hougaard, D. M., Christensen, E., Gahrn, B., Christensen, M., Bross, P., Vested, A., Simonsen, H., Skogstrand, K., Olpin, S., Brandt, N. J., Skovby, F., Nørgaard-Pedersen, B., & Gregersen, N. (2012). MCAD deficiency in Denmark. *Molecular genetics and metabolism*, 106(2), 175-188. <https://doi.org/10.1016/j.ymgme.2012.03.018>
8. NxGen MDx (2021) Data on file.