Identifying family risk for Danon disease with family variant testing

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Danon disease is a rare, X-linked genetic disorder caused by mutations in the *LAMP2* gene.[1-3] If your patient has been diagnosed with Danon disease, understanding their family's genetic risk is essential. **Family variant testing can help identify who else may be at risk, providing them with knowledge, direction, and tools as they take the first steps toward treatment.**[1]

If a *LAMP2* mutation is identified, who else should get tested?

Do you have a patient who has tested positive for Danon disease? If so, international guidelines suggest the rest of their family should also be tested.

Even if they don't show symptoms of Danon disease, they may carry the *LAMP2* mutation.[2]

Early identification is key![1]

Family members who may be at risk include[1]:

- Parents
- Siblings
- Children
- Aunts/uncles/cousins
- Grandparents

Why is family variant testing important?

By identifying family members who carry the *LAMP2* mutation, it's possible to:

- Detect Danon disease and begin monitoring early[2]
- Provide timely medical interventions[2]
- Empower families with knowledge and opportunities to participate in clinical trials[3]

How can family variant testing help with Danon disease?

Danon disease can cause health problems such as:

- Severe heart disease (cardiomyopathy), often leading to heart failure that requires transplantation[2,3]
- Muscle weakness[2,3]
- Cognitive impairment[2,3]
- Vision issues[2,3]

Early diagnosis allows for:

- Specialized monitoring and care[1]
- Family planning support[1]

TAKE ACTION TODAY! Flip over to learn how you can order testing for your patients and their families.

Rocket Pharma offers free genetic counseling and testing through the Mission: Genome program*

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If your patient receives a positive test for Danon disease, you can order family variant testing to determine if others could be at risk.

Contact Mission: Genome	Scan the QR code below to visit **MissionGenome.com**.
Order genetic testing	Place the order for genetic testing for appropriate family members and discuss next steps with them.
Receive results	You and your patients will receive secure, personalized results with recommendations for follow-up care.

Why choose Mission: Genome?

- No-cost genetic counseling and testing*
- Personalized, supportive care
- Comprehensive follow-up resources

READY TO GET STARTED? Order counseling or testing for your patients and their families at **MissionGenome.com**—counseling appointments are available within 1 week!

If the family would like genetic counseling prior to testing, have them follow these steps:

1. Visit **MissionGenome.com** and select Genetic Counseling
2. On the Genome Medical page, select Schedule Now
3. Schedule a counseling consultation
4. After the consultation, the counselor will assess whether family testing is recommended
5. Counselor will order the appropriate test for family members

Even if all members of the family are not your patients, **Mission: Genome** will provide genetic counseling and testing at no cost* for eligible patients.

*For eligible patients and their families. Eligibility requirements can be found on testing providers' web pages.

References: 1. Wilde AAM, Semsarian C, Márquez MF, et al. European Heart Rhythm Association (EHRA)/Heart Rhythm Society (HRS)/Asia Pacific Heart Rhythm Society (APHRS)/Latin American Heart Rhythm Society (LAHRS) expert consensus statement on the state of genetic testing for cardiac diseases. *Europace*. 2022;24(8):1307-1367. **2.** Hong KN, Eshraghian EA, Arad M, et al. International consensus on differential diagnosis and management of patients with Danon disease: JACC state-of-the-art review. *J Am Coll Cardiol*. 2023;82(16):1628-1647. **3.** Brambatti M, Caspi O, Maolo A, et al. Danon disease: gender differences in presentation and outcomes. *Int J Cardiol*. 2019;286:92-98.

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