

Disease

# Glycogen storage disease, type IXd<sup>1</sup>

Gene

## PHKA1

|                                          |                                                                                                                                                                                                                                                                             |
|------------------------------------------|-----------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| Inheritance                              | XL                                                                                                                                                                                                                                                                          |
| Current Treatments                       | Avoid vigorous exercise; medications like succinylcholine and statins that can cause rhabdomyolysis.                                                                                                                                                                        |
| Evidence for Treatment                   | guideline <sup>2</sup>                                                                                                                                                                                                                                                      |
| Non-Molecular Testing Availability       | no                                                                                                                                                                                                                                                                          |
| Non-Molecular Test                       |                                                                                                                                                                                                                                                                             |
| Genes Included in Disease Frequency      | PHKA1, PHKA2, PHKB, PHKG2                                                                                                                                                                                                                                                   |
| Experimental / Investigational Therapies | <a href="https://clinicaltrials.gov/ct2/results?cond=Glycogen+Storage+Disease+IX&amp;term=&amp;cntry=&amp;state=&amp;city=&amp;disease=">https://clinicaltrials.gov/ct2/results?cond=Glycogen+Storage+Disease+IX&amp;term=&amp;cntry=&amp;state=&amp;city=&amp;disease=</a> |

References

<sup>1</sup> <https://www.ncbi.nlm.nih.gov/books/NBK55061/#gsd9.Summary>

<sup>2</sup> <https://www.ncbi.nlm.nih.gov/pubmed/30659246>