# DISEASE:
# FKRP-related limb-girdle muscular dystrophy R9

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| **NAME:** | FKRP-related limb-girdle muscular dystrophy R9 |
| **DESCRIPTION:** | A form of autosomal recessive limb-girdle muscular dystrophy that presents a highly variable age of onset and phenotypic spectrum typically characterized by slowly progressive proximal weakness of the pelvic and shoulder girdle musculature (predominantly affecting the lower limbs), frequently associated with waddling gait, scapular winging, calf and tongue hypertrophy, exercise-induced myalgia, abdominal muscle weakness, cardiomyopathy, respiratory muscle involvement, and myoglobinuria and/or elevated creatine kinase serum levels. |
| **ORPHACODE:** | 34515 |
| **SYNONYMS:** | Autosomal recessive limb-girdle muscular dystrophy type 2I<br>FKRP-related LGMD R9<br>LGMD due to FKRP deficiency<br>LGMD type 2I<br>LGMD2I<br>Limb-girdle muscular dystrophy due to FKRP deficiency<br>Limb-girdle muscular dystrophy type 2I |
| **XREF(S):** | Orphanet<br>ICD-10<br>OMIM |

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| **ANALYTE(S):** | FKRP |
| **CREATED:** | 13 May 2019 - 01:02 |
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# RELATED CONTENT

## Related Genetic Tests

- Cardiomyopathy, hereditary (gene panel)

## Related Laboratories

- Centrum Medische Genetica - UZ Brussel VUB

## Related Analytes

- fukutin related protein

## Related Gene Panels

- Cardiomyopathy, hereditary (208 genes) - VUB