Type ix glycogenosis (BRESOLIN disease) - **phosphoglycerate kinase** deficiency.

* X-linked inheritance.

KLINIKA:

1. **Seizures**, **mental retardation**.
2. Severe **hemolytic anemia**
3. Infrequent **dynamic myopathy** (exercise intolerance, myoglobinuria). see Met1 p.

Type x glycogenosis (TONIN disease) - **phosphoglycerate mutase** deficiency.

KLINIKA – **dynamic myopathy** (exercise intolerance, myoglobinuria). see Met1 p.

Type xI glycogenosis (TSUJIMO disease) - **lactate dehydrogenase** deficiency.

KLINIKA:

1. **Dynamic myopathy** (exercise intolerance, myoglobinuria). see Met1 p.
2. **Rash** (M-lactate dehydrogenase is dominant form of enzyme expressed in skin).

DIAGNOSTIKA - forearm exercise test: no [lactate] rise, but normal [pyruvate] rise.

Satoyoshiy disease - **phosphohexose isomerase** (glucose-6-P ↔ fructose-6-P) deficiency.

KLINIKA:

* 1. Chronic nonspherocytic **hemolytic anemia**
  2. **Dynamic myopathy** (exercise intolerance, myoglobinuria). see Met1 p.
* exercise tolerance is present in childhood, but myopathy worsens later in life.
* muscle contractures do not occur.

Thomson disease – **phosphoglucomutase** (glucose-1-P ↔ glucose-6-P) deficiency.

One patient:

* 1. Numerous episodes of **supraventricular tachycardia**
  2. Mild static **weakness** and poor muscle development (no exercise intolerance).
  3. Bulky and firm calf muscles, **shortened Achilles' tendons** (toe-walking)

Another patient - profound **metabolic acidosis**, recurrent **vomiting**, lethargy, **poor weight gain**.

Glycogenoses – additional info

Pompe disease type 2 glycogenosis

FDA approved Lumizyme® (alglucosidase alfa) for patients ≥ 8 years with late-onset (non-infantile) Pompe disease.