

# **GSD III**

**Milton Keynes 2005**

**G.P.A.Smit UMC Groningen NL**

# GSD III

- What do we know
- What should we know
- How can we achieve that

# Glycogen Storage Disease Type III

Clinical presentation:

Short stature

Protruded abdomen

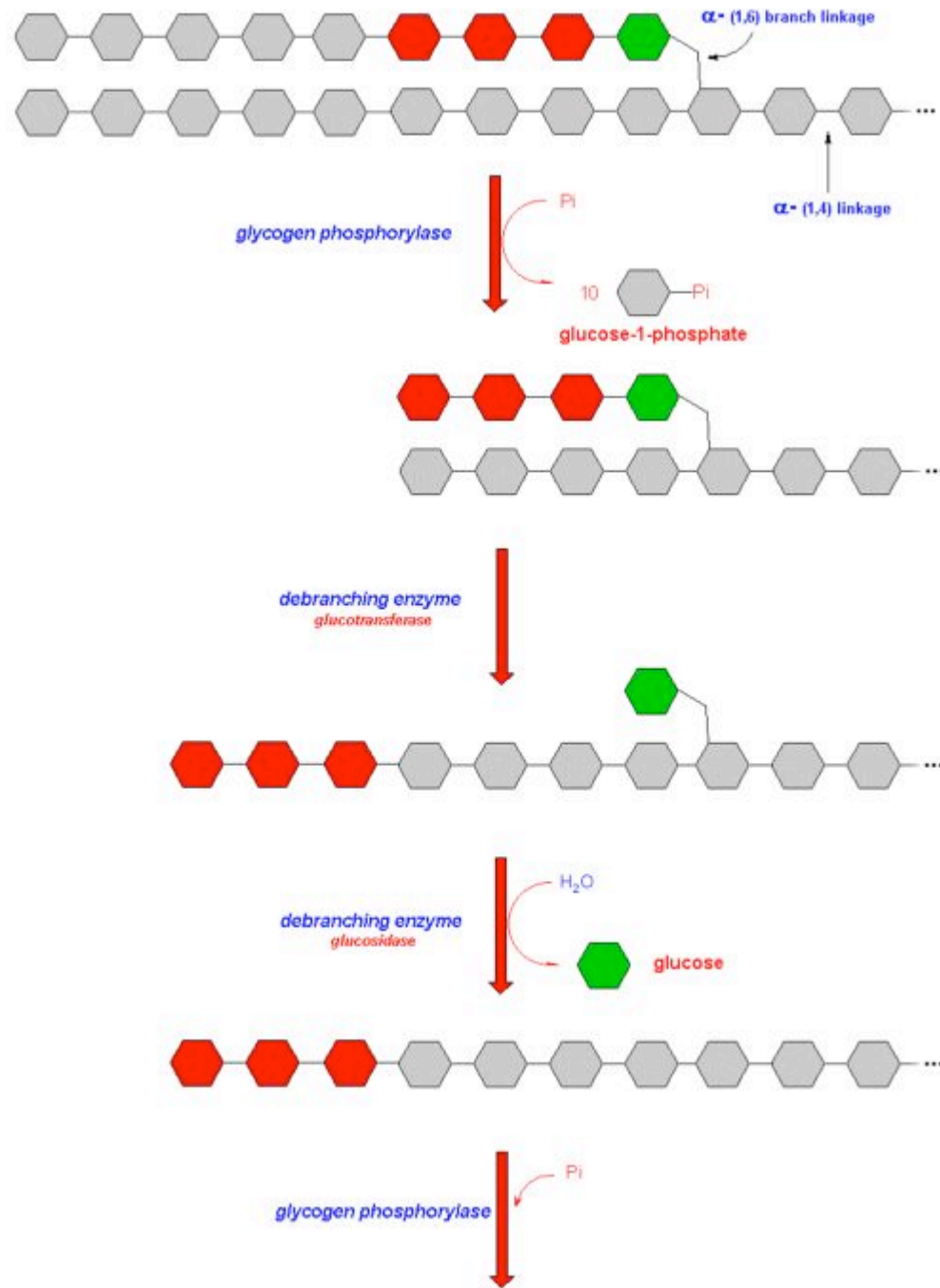
Truncal obesity

hypotonia

Autosomal recessive 1p21

# Glycogen Storage Diseases

- Type I            Ia            Ib
- Type III
- Type IV
- Type Phosphorylase deficiency
  
- Liver related



# Glycogen Storage Disease Type III

# GSD III Subtypes

- IIIa liver, muscle, heart, peripheral nerves
- III b liver
- IIIc very rare glucosidase only
- III d very rare transferase only

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# Glycogen Storage Disease Type III

Metabolic derangements:

Hypoglycemia

Hyperketonemia

Hyperlipidemia

Hyperuricemia

# Glycogen Storage Disease Type III

Therapy:

Frequent meals

Carbohydrate enriched

    Nocturnal gastric drip feeding

    Complex carbohydrates

Protein enriched

# Glycogen Storage Disease Type III

Complications:

Growth

Cirrhosis

Hepatocellular carcinoma

Myopathy, Cardiomyopathy

Peripheral nerves

Diabetes mellitus

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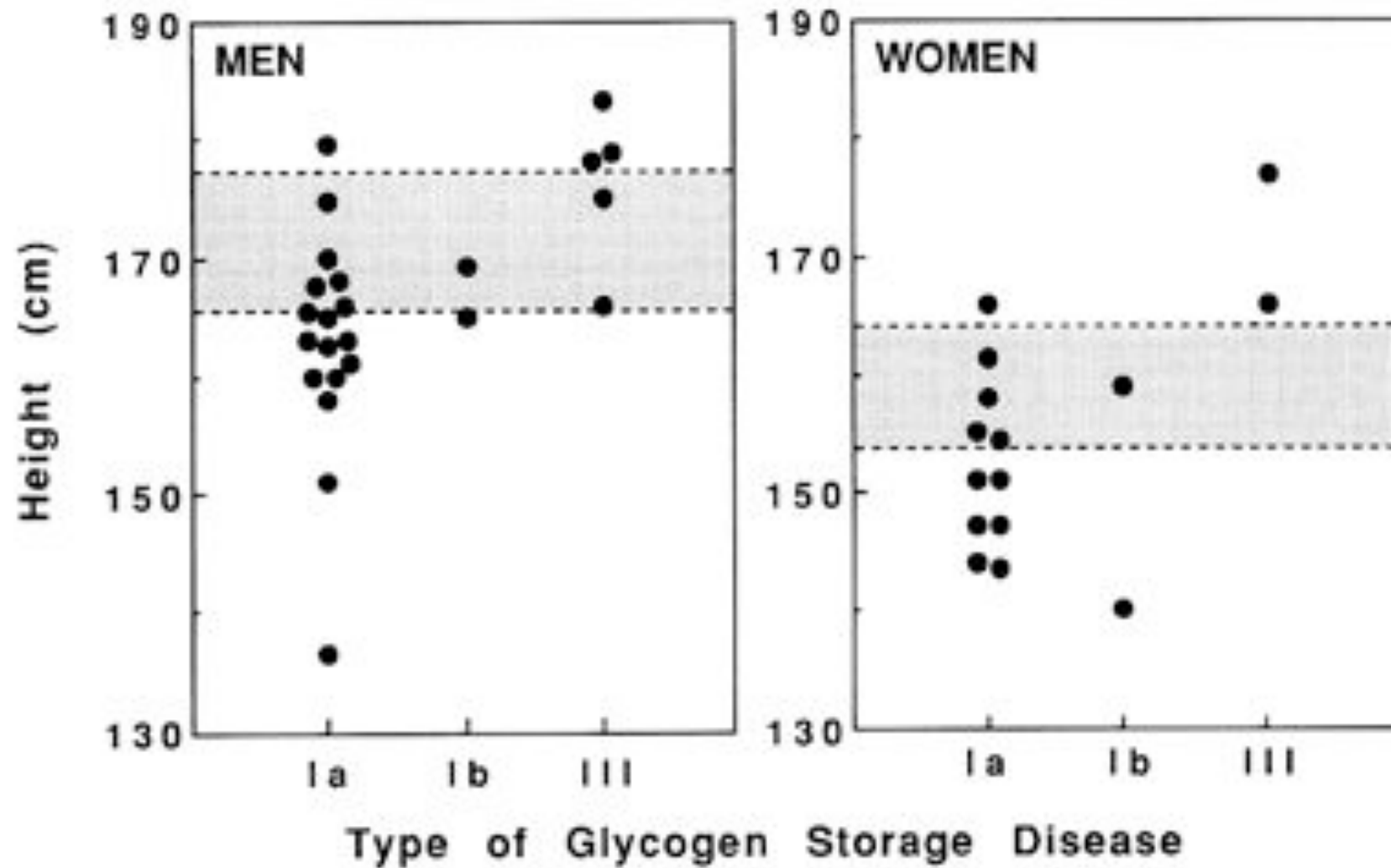
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Talente et al 1994

**GSD I**

**Metabolic control?**

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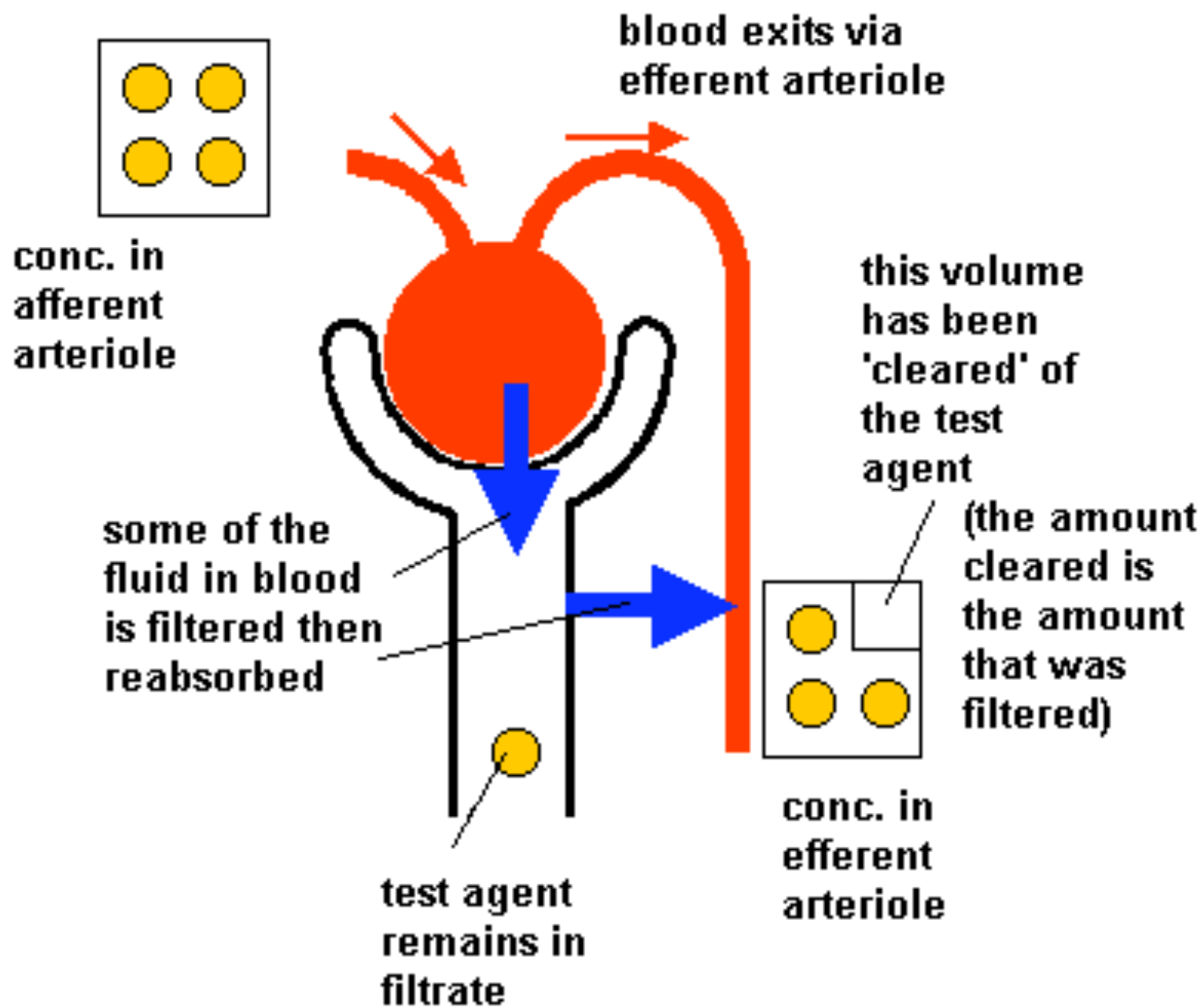
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**Metabolic control?**

**GSD I**



**Metabolic control?**

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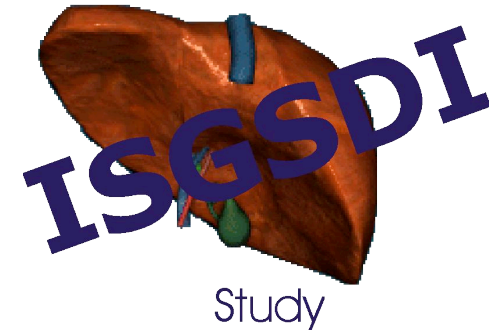
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**Metabolic control?**

**GSD I**

# Recommendations ESGSD I Study



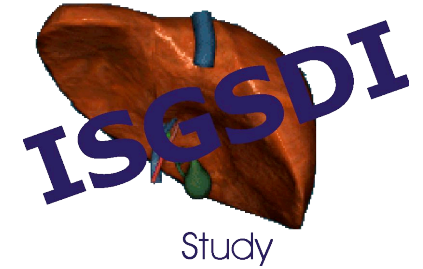
- Pre-prandial glucose  $> 3,5-4,0$  mmol/l
- Urine lactate  $< 0,6$  mmol/l or  $< 0,06$  mmol/mmol creatinine
- Serum uric acid in high normal range
- Venous BE  $> -5$  mmol/l
- Venous bicarbonate  $> 20$  mmol/l
- Serum TG concentration  $< 6,0$  mmol/l
- BMI between 0.0 and + 2.0 SDS

ISGSD I

Collaborative

study

ISGSD III ?



# ISGSD I Study

21 metabolic centres from 14 countries

- Austria
- Belgium
- Czech Republik
- France
- Germany
- **Great Britain**
- Hungary
- Italy
- Israel
- The Netherlands
- Poland
- Sweden
- Turkey
- United States of America
- $\pm$  500 patients expected

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