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Glycogen storage disease type I

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Glycogen Storage Disease type I: clinical, biochemical and genetic aspects, and implications for treatment and follow-up.

(management of glycogen storage disease type I)

Jan Peter Rake



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RIJKSUNIVERSITEIT GRONINGEN

**Glycogen Storage Disease type I:
clinical, biochemical and genetic
aspects, and implications for
treatment and follow-up.**

(management of glycogen storage disease type I)

Proefschrift

ter verkrijging van het doctoraat in de
Medische Wetenschappen
aan de Rijksuniversiteit Groningen
op gezag van de
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in het openbaar te verdedigen op
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om 16.00 uur

door

Jan Peter Rake

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te Groningen

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'Toutes les expériences s'enchaînent naturellement pour établir que le sucre, véritable produit d'une sécrétion intérieure, à laquelle j'ai donné le nom de glycogénie, prend naissance dans le foie aux dépens des éléments du sang et indépendamment de l'alimentation féculente et sucrée'

Claude Bernard, 24-03-1855, Vingt-cinquième leçon: 'Sur la glycogénie animale'

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abbreviations

| | |
|-------------------------|---|
| ACE | angiotensin converting enzyme |
| BMC | bone mineral content |
| BMD | bone mineral density |
| BMD _{areal} | areal bone mineral density in g/cm ² |
| BMD _{bone age} | areal bone mineral density corrected for delayed bone maturation in g/cm ² |
| BMD _{vol} | volumetric bone mineral density in g/cm ³ |
| BMI | body mass index |
| CD | Crohn's disease |
| CNGDF | continuous nocturnal gastric drip feeding |
| CVS | chorionic villus samples |
| DXA | dual energy X-ray absorptiometry |
| E/A ratio | early filling velocity / atrial filling velocity |
| ER | endoplasmic reticulum |
| ERPF | effective renal plasma flow |
| ESGSD I | European study on glycogen storage disease type I |
| FMs | frequent meals |
| G6P | glucose-6-phosphate |
| G6Pase | glucose-6-phosphatase |
| G6PC | glucose-6-phosphatase gene |
| G6PT | glucose-6-phosphate translocase gene |
| GCSF | granulocyte colony stimulating factor |
| GFR | glomerular filtration rate |
| GLUT | glucose transporter |
| GSD(s) | glycogen storage disease(s) |
| HDL | high density lipoprotein |
| IBD | inflammatory bowel disease |
| IDDM | insulin dependent diabetes mellitus |
| IMT | intima-media thickness |
| ISGSD I | international study on glycogen storage disease type I |
| KT | kidney transplantation |
| LDL | low density lipoprotein |
| LT | liver transplantation |
| LVM(I) | left ventricular mass (index) |
| MD | metabolic derangement |
| MIDA | mass isotopomer distribution analysis |
| MUFA | monounsaturated fatty acids |
| PET | positron emission tomography |
| PCOs | polycystic ovaries |
| PCR | polymerase chain reaction |
| PLR | partial liver resection |
| PUFA | polyunsaturated fatty acids |
| RRT | renal replacement therapy |
| RWT | relative wall thickness |
| SDS | standard deviation score |
| SFA | saturated fatty acids |
| SSCP | single strand conformational polymorphism |
| UCCS | uncooked (corn) starch |
| UK | United Kingdom |
| USA | United States of America |
| VLDL | very low density lipoprotein |
| XO | xanthine oxidase |