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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
Rector Magnificus, dr. F. Zwarts,
in het openbaar te verdedigen op
woensdag 8 oktober 2003
om 16.00 uur

door

Jan Peter Rake

geboren op 21 augustus 1968
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