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Regulation and modulation of growth : insights from human and animal studies

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Curriculum vitae

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The author of this thesis was born on October 13th, 1974 in Uitgeest, the Netherlands. She attended secondary school at the 'Gymnasium Felisenum' in Velsen-Zuid and passed her exam in 1993. From 1993-2001, she studied Medical Biology at the University of Amsterdam. During her study, she performed two research projects at the Department of Pediatrics of the Academic Medical Center Amsterdam (AMC) on human gallbladder mucin (Dept. of Pediatric Gastroenterology and Nutrition; Prof. dr. H.A. Büller and dr. A. Einerhand), and on carnitin biosynthesis (Dept. of Genetic Metabolic Disorders; Prof. dr. R. Wanders and dr. R. Ofman). In September 2001, she received her Medical Biology degree (*cum laude*). From 1998-2001 she studied Medicine at the University of Amsterdam from 1998-2001 and received a Scholarship for Excellent Students on a double degree programme (Medicine and Medical Biology). She obtained her medical degree in September 2003 and subsequently worked for 6 months as a pediatric resident at the Zaandam Medical Center (Mr. M. Westra). In April 2004, she started as a research fellow at the Department of Pediatrics of the Leiden University Medical Center (Prof. dr. J.M. Wit, dr. M. Karperien and dr. W. Oostdijk). Her application for an AIOSKO program sponsored by NWO (the Netherlands Organization for Health Research and Development), a combination of pediatric residency and PhD studentship, was honored. During her research period, she performed working visits to the Instituto Cajal in Madrid, Spain (Prof. dr. L.M. Garcia-Segura and Dr. S. Veiga), the University of Veterinary Medicine (Prof. Böck) and the Pediatric Department of the University Hospital of Vienna, Austria (Prof. dr. G. Haeusler). In July 2009, she started her clinical training in pediatrics at the Leiden University Medical Center (Prof. dr. H.A. Delemarre-van de Waal). As of January 2010, she works at the Juliana Children's Hospital in The Hague, the Netherlands (Dr. F. Brus), to fulfill the non-academic part of her training in pediatrics.



List of publications

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Van Gool SA, Wit JM, de Schutter T, de Clerck N, Postnov AA, Kremer Hovinga S, van Doorn J, Veiga SJ, Garcia-Segura LM, Karperien M. Impaired body weight and tail length gain after treatment with the aromatase inhibitor exemestane in male rats. *Horm Res Paediatr* 2010;73(5):376-85.

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Emons JAM, Decker E, Yu X, Pfirzer H, van Gool SA, Chagin A, Savendahl L, Gretz N, Wit JM, Rappold G, Karperien M. Genome-wide screening of two human growth plates during pubertal development. Submitted for publication.



List of abbreviations

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AH	Adult height
AI	Aromatase inhibitor
ALS	Acid-labile subunit
ANOVA	Analysis of variance
AR(KO)	Androgen receptor (knockout mouse)
ArKO	Aromatase knockout mouse
BA	Bone age
BM(A)D	Bone mineral (apparent) density
BMI	Body mass index
BMP	Bone morphogenetic protein
CA	Chronological age
CAH	Congenital adrenal hyperplasia
CPP	Central precocious puberty
DHT	Dihydrotestosterone
DSD	Disorder of sex development
DXA	Dual energy x-ray absorptiometry
E	Exemestane
ECM	Extracellular matrix
ER($\alpha/\beta/\alpha\beta$ KO)	Estrogen receptor (alpha/beta/alpha+beta knockout mouse)
FH	Final height
FSS	Familial short stature
GF	Growth factor
(h)GH	(human) Growth hormone
GHR	Growth hormone receptor
GHRH	Growth hormone releasing hormone
GHD	Growth hormone deficiency
GnRH _a	Gonadotropin releasing hormone agonist
GWAS	Genome-wide association studies
H	Height
H&E	Haematoxylin and Eosin
IGF-I	Insulin-like growth factor I
IGF1R	Insulin-like growth factor receptor
IGFBP3	IGF binding protein 3

ISS	Idiopathic short stature
KEGG	Kyoto encyclopedia of genes and genomes
LL	Leg length
MAS	McCune-Albright syndrome
micro-CT	X-ray microtomography
(hf)MSC(s)	(human fetal) Mesenchymal stem cell(s)
NFSS	Non-familial short stature
NHANES	National health and nutrition examination survey
ORCHX	Orchidectomy
OVX	Ovariectomy
PAH	Predicted adult height
PBI	Pediatric bone index
PCA	Principal component analysis
PCO	Polycystic ovaries syndrome
qPCR	Real-time quantitative polymerase chain reaction
P/H	Proliferative zone/hypertrophic zone ratio
PICP	Procollagen type IC propeptide
PIIINP	Procollagen type III N propeptide
PI3K	Phosphoinositide-3-kinase
PLC	Placebo
PTHrP	Parathyroid hormone related protein
RCT	Randomized controlled clinical trial
SD(S)	Standard deviation (score)
SERM	Selective estrogen receptor modulator
SGA	Short for gestational age
SH	Sitting height
SNP	Single nucleotide polymorphism
TBW	Total body weight
TGF	Transforming growth factor
(c)TH	(conditional) Target height
Wnt	Wingless-type MMTV integration site family