



Research Annual Report 2011



Medical Genetics Center (MGC)

Molecular and cellular basis of genome instability in cancer

Bioinformatics

Gene-expression during embryonal development

Gene-therapy

Chromatin regulation in development and disease

Forensic molecular biology

Clinical and experimental aspects of embryogenesis and early placental development

Development disorders and congenital malformations

Reproduction and development

Identification and characterisation of disease genes

EMC MGC-01-12-03 - Molecular and cellular basis of genome instability in cancer

Programme (brief description)

Preserving the genome is of prime importance to all living systems. However, the integrity of DNA, the carrier of genetic information, is continuously threatened by endogenous and exogenous agents and by intrinsic instability of chemical bonds in DNA itself. Oxidative stress, in part derived from the normal oxidative phosphorylation UV- and X-rays and numerous chemicals induce a wide variety of lesions in DNA. Obviously, this affects proper functioning of the DNA and can lead to cell death, cancer, inborn disorders, and overall functional decline contributing to ageing. To counteract the gradual erosion of the vital genetic information and prevent its pleiotropic detrimental consequences an intricate network of genome care-taking and protection systems has evolved. DNA repair pathways and cell cycle control mechanisms constitute an important component of this genome protection network. The overall objective of this program is to understand the molecular mechanism and the biological impact of the systems designed to preserve the vital genetic material.

Our primary objectives are:

1. Understanding of the molecular mechanisms, specificity, fidelity and in vivo functioning of major multi-enzyme repair and damage-response pathways: nucleotide excision repair (NER), double strand break (dsb) repair and replication damage tolerance mechanisms, as well as damage-induced cell cycle arrest.
2. Elucidation of the molecular defects of genetic disorders with system deficiencies in DNA repair and other genome maintenance systems. Examples are NER disorders, xeroderma pigmentosum (XP), Cockayne's syndrome and trichothiodystrophy. These diseases are characterized by sun (UV) sensitivity, frequently neurological abnormalities, poor development, frequently features of premature ageing or in case of XP predisposition to skin cancer. Similarly, we wish to identify and/or understand other human conditions associated with impaired dsb repair, damage tolerance, or cell cycle response.
3. Generation of mouse models for human NER, dsb repair and damage tolerance deficiencies to assess the biological impact of these systems. The approaches followed include isolation and functional characterization of mammalian repair genes and proteins, analysis of these proteins in vitro and in living cells, their involvement in human repair syndromes, their use for inducing specific repair defects in the mouse germ-line by gene targeting as well as analysis of defects in these systems at the level of mRNA expression (genomics) and proteins (proteomics). Insight into DNA repair mechanisms is relevant for understanding the effect of DNA damage on vital processes as transcription, replication, recombination and cell cycle progression. Indirectly this process affects the origin of inborn defects, carcinogenesis, genome stability and ageing. Finally, a new line of research is aimed at elucidating the intriguing molecular mechanism and biological impact of the circadian clock in mammals.

Key Figures

| <i>Department</i> | <i>Sp1</i> | <i>Sp2</i> | <i>Sp3</i> | <i>Sp4</i> | <i>SpTot</i> | <i>T1</i> | <i>T2</i> | <i>T3</i> | <i>Ttot</i> | <i>IS</i> | <i>FS</i> | <i>OP</i> | <i>Ptot</i> |
|-------------------|--------------|--------------|-------------|-------------|--------------|-----------|-----------|-----------|-------------|-----------|-----------|-----------|-------------|
| genetics | 11,55 | 36,54 | 6,08 | 0,67 | 54,84 | | | | | | | | |
| Total | 11,55 | 36,54 | 6,08 | 0,67 | 54,84 | 2 | 2 | 0 | 4 | 52 | 5 | 0 | 61 |

Thesis

Brand, K. (2011, juni 22). *Transcripts from the Circadian Clock & Telling time and Season*. EUR (169 pag.). Prom./coprom.: Prof. Dr. G.T.J. van der Horst & Prof. Dr. J.H.J. Hoeijmakers.

Faesen, AC (2011, november 09). *Activity modulation of ubiquitin specific proteases*. EUR. Prom./coprom.: Prof. Dr. T.K. Sixma.

Jagalur Basheer, N. (2011, mei 18). *Role of Oct6 in Peripheral Nerve Myelination*. EUR (160 pag.). Prom./coprom.: Prof. Dr. Ir. D.N. Meijer.

Vargas, M.P.A. (2011, november 09). *Structural and functional studies on the ubiquitin-specific protease family*. EUR. Prom./coprom.: Prof. Dr. T.K. Sixma.

Article/Letter to the editor

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Part of book - abstract

Eppink, B., Essers, J. & Kanaar, R. (2011). Interplay and quality control of DNA damage repair mechanisms. In K. Rippe (Ed.), *Genome Organization and Function in the Cell Nucleus* (pp. 395-415). Weinheim, Germany: Wiley-VCH.

Pothof, J. & Gent, D.C. van (2011). Spatiotemporal aspects of MicroRNA mediated gene regulation. In L.J. Collins (Ed.), *RNA infrastructure and networks*. Austin TX, USA: Landes Bioscience and Springer.

EMC MGC-02-02-01 – Bioinformatics

Programme (brief description)

In collaboration with other departments the department of Bioinformatics multidisciplinary team supports projects that generate genomics and proteomics data from basis research, forensics studies, molecular diagnostics and clinical trials.

The center also runs a research program of its own, which provides the biological and technological basis of all the other activities. It concentrates on the way the genome as a whole contributes to the evolution, development, structure and function of the brain. Among others it involves analysis of gene expression in cells of the brain and combines genomics, proteomics and cytogenetic data to identify genes associated with neurological disorders.

The Erasmus MC Bioinformatics department initiated a translational medicine program. This effort will assist in the critical task of moving medical research closer to commercial Ready medical technology that can be applied within and outside Erasmus University Medical Center. Moving medicine forward requires data integration from Bench to Bedside and from Patient to Population. The bioinformatics department plays a central role in linking research data onto clinical data using state of the art ICT technology and medical informatics expertise. The ultimate goal is to identify biomarkers linking genotypic data and phenotypic data to support processes such as determination of genetic risk, patient stratification, disease staging, treatment selection and evaluation of outcome to improve the quality of life of the patient. This strategy will also provide insight in environmental factors, lifestyle Information, and treatment history that correlate with the natural history of the disease.

Key Figures

| Department | Sp1 | Sp2 | Sp3 | Sp4 | SpTot | T1 | T2 | T3 | Ttot | IS | FS | OP | Ptot |
|-------------------|-------------|------------|------------|------------|--------------|-----------|-----------|-----------|-------------|-----------|-----------|-----------|-------------|
| bioinformatics | 5,17 | 1 | | 1,5 | 7,67 | | | | | | | | |
| Total | 5,17 | 1 | 0 | 1,5 | 7,67 | 1 | 0 | 0 | 1 | 18 | 0 | 0 | 19 |

Thesis

Rousian, M.. *Embryonic Development in Virtual Reality*. EUR. Prom./coprom.: Prof. Dr. E.A.P. Steegers & Dr. N. Exalto.

Article/Letter to the editor

Bralten, L.B., Nouwens, S.C.A., Kockx, C.E.M., Erdem, L., Hoogenraad, C.C., Kros, J.M., Moorhouse, M.J., Sillevs Smitt, P.A.E., Spek, P.J. van der, Ijcken, W.F.J. van, Stubbs, A.P. & French, P.J. (2011). Absence of Common Somatic Alterations in Genes on 1p and 19q in Oligodendrogliomas. *PLoS One*, 6(7).

Bralten, L.B.C., Kloosterhof, N.K., Balvers, R, Sacchetti, A., Lapre, L, Lamfers, M., Leenstra, S., Jonge, H.R. de, Kros, J.M., Jansen, E.E.W., Struys, EA, Jakobs, C., Salomons, G.S., Diks, SH, Peppelenbosch, M, Kremer, A., Hoogenraad, C.C., Sillevs Smitt, P.A.E. & French, P.J. (2011). IDH1 R132H Decreases Proliferation of Glioma Cell Lines In Vitro and In Vivo. *Annals of Neurology*, 69(3), 455-463.

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Rousian, M., Koning, A.H.J., Spek, P.J. van der, Steegers, E.A.P. & Exalto, N. (2011). Virtual reality for embryonic measurements requiring depth perception. *Fertility & Sterility*, 95(2), 773-774.

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EMC MGC-02-13-02 - Gene-expression during embryonal development

Programme (brief description)

Research is aimed at two main interests; the development of the hemopoietic system and the development of the brain including the neural crest. Study of the hemopoietic system is focussed on two different aspects, the control of gene expression during development and differentiation and on the processes that underlie the induction of the hematopoietic at different times of development. The control of gene expression is mainly focussed on the regulation of transcription of the globin genes and the transcription factors involved in this process. We are also very interested in B cell development, a cell type that responds to an ordered set of cell surface signals for its development resulting in the rearrangement of immunoglobulin genes. These studies are focused on one of the genes involved in signalling and the genes that are responsible for the rearrangement process. Hemopoietic induction is mainly focussed on the characterization of early stem cells that form the foetal/adult blood system. The study of the development of neural crest cells and the brain is focused on three separate areas. Firstly, we are interested in the molecular controls underlying Schwann cell development, secondly the molecular defects that underlie DiGeorge's syndrome and thirdly the relationship between cellular structure and function in the brain. Included in the last programme is the elucidation of the molecular defects in Alzheimer's disease.

Key Figures

| Department | Sp1 | Sp2 | Sp3 | Sp4 | SpTot | T1 | T2 | T3 | Ttot | IS | FS | OP | Ptot |
|-------------------|-------------|--------------|-------------|-------------|--------------|-----------|-----------|-----------|-------------|-----------|-----------|-----------|-------------|
| cell biology | 15,2 | 23,34 | 2,92 | 1,89 | 43,35 | | | | | | | | |
| genetics | 0,5 | 0,5 | | | 1 | | | | | | | | |
| Total | 15,7 | 23,84 | 2,92 | 1,89 | 44,35 | 6 | 0 | 1 | 7 | 18 | 1 | 0 | 26 |

Thesis

Esteghamat Hanzaei, F. (2011, oktober 20). *Erythropoiesis and Hemoglobin Regulation: A journey from laboratory to disorders*. EUR (212 pag.). Prom./coprom.: Prof. Dr. F.G. Grosveld & Prof. Dr. J.N.J. Philipsen.

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Article/Letter to the editor

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EMC MGC-02-13-03 - Gene-therapy

Programme (brief description)

Research is centered towards some fundamental mechanisms which determine the development of disease specific gene therapies. The diseases under study are: cystic fibrosis, thalassemia, immunodeficiencies, Crigler Najjar, neoplasia and restenosis. The present and future research is focused on the development of non viral delivery vehicles with particular attention to the process of the transport of DNA into the nucleus in non dividing cells and the integration of DNA into the host genome. In addition we are developing replicating vectors based on papilloma virus replication origins. Part of this project is a joint effort with the department of Pediatrics (ZKL 540207), Cardiology (TOC 430604) and Surgery (HKG 470101).

Key Figures

| Department | Sp1 | Sp2 | Sp3 | Sp4 | SpTot | T1 | T2 | T3 | Ttot | IS | FS | OP | Ptot |
|-------------------|-------------|-------------|------------|------------|--------------|-----------|-----------|-----------|-------------|-----------|-----------|-----------|-------------|
| cell biology | 7,95 | 4,78 | 1 | 3,6 | 17,33 | | | | | | | | |
| Total | 7,95 | 4,78 | 1 | 3,6 | 17,33 | 1 | 0 | 0 | 1 | 15 | 2 | 0 | 18 |

Thesis

Buijs-Offerman, R.M.G.B. (2011, juni 22). *Studies on Airway Inflammation and Remodeling in a Cystic Fibrosis F508del FCTR Mouse Model*. EUR (202 pag.). Prom./coprom.: Prof. Dr. F.G. Grosveld.

Article/Letter to the editor

Berge, D. ten, Kurek, D.M., Blauwkamp, T, Koole, W, Maas, A., Eroglu, E, Siu, RK & Nusse, R (2011). Embryonic stem cells require Wnt proteins to prevent differentiation to epiblast stem cells. *Nature Cell Biology*, 13(9), 1070-U88.

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Lunardi, A., Gaboli, M., Giorgio, M., Rivi, R., Bygrave, A., Antoniou, M., Drabek, D., Dzierzak, E.A., Fagioli, M., Salmena, L., Botto, M, Cordon-Cardo, C., Luzzatto, L., Pelicci, P.G., Grosveld, F.G. & Pandolfi, P.P. (2011). A Role for PML in Innate Immunity. *Genes & Cancer*, 2(1), 10-19.

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Robin, C.I., Ottersbach, K., Boisset, J.C., Oziemlak, A. & Dzierzak, E.A. (2011). CD41 is developmentally regulated and differentially expressed on mouse hematopoietic stem cells. *Blood*, 117(19), 5088-5091.

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Wilke, M., Buijs-Offerman, R.M.G.B., Aarbiou, J., Colledge, W.H., Sheppard, DN, Touqui, L, Bot, A.G.M., Jorna, H.J.J., Jonge, H.R. de & Scholte, B.J. (2011). Mouse models of cystic fibrosis: Phenotypic analysis and research applications. *Journal of Cystic Fibrosis*, 10, S152-S171.

Yokomizo, T., Ng, CEL, Osato, M & Dzierzak, E.A. (2011). Three-dimensional imaging of whole midgestation murine embryos shows an intravascular localization for all hematopoietic clusters. *Blood*, 117(23), 6132-6134.

EMC MGC-02-21-01 - Chromatin regulation in development and disease

Programme (brief description)

The goal of this research program is to understand the mechanism of gene expression control during development and disease. We are interested in how the expression of the eukaryotic genome is regulated. The Verrijzer group focusses on the role of chromatin regulation in development and disease. Over the last decade or so, it has become clear that chromatin structure forms an integral part of the mechanisms by which gene transcription is controlled in eukaryotic cells. Their studies focus on three related topics: (1) epigenetic control of transcription regulation in (stem) cell differentiation and cancer. (2) Transcription control by protein (de)ubiquitylation. (3) gene control by metabolic enzymes. The Mahmoudi group explores the molecular mechanisms that regulate gene expression with an emphasis on identification of specific molecular targets in two disease states: colorectal cancer and latent HIV infection. Their research aims to (1) identify molecular targets in CRC therapy and (2) study and manipulate the molecular mechanisms of HIV latency establishment and re-activation. Research in the Fornerod group aims to gain understanding of the role of the nuclear envelope in development and disease. The nuclear envelope separates the nucleus and cytoplasm of and is one of the most important borders within the eukaryotic cell. A main focus of the current research is on the role of nucleoplasmic nucleoporins expressed as a consequence of leukemia-associated chromosome translocations. Finally, Dr. Demmers heads the EMC proteomics centre. His own research centers on proteomic analysis of cell differentiation dynamics of normal and diseased cells.

Key Figures

| <i>Department</i> | <i>Sp1</i> | <i>Sp2</i> | <i>Sp3</i> | <i>Sp4</i> | <i>SpTot</i> | <i>T1</i> | <i>T2</i> | <i>T3</i> | <i>Ttot</i> | <i>IS</i> | <i>FS</i> | <i>OP</i> | <i>Ptot</i> |
|-------------------|--------------|-------------|------------|------------|--------------|-----------|-----------|-----------|-------------|-----------|-----------|-----------|-------------|
| biochemistry | 11,29 | 5,08 | | 1 | 17,37 | | | | | | | | |
| Total | 11,29 | 5,08 | | 1 | 17,37 | 0 | 0 | 0 | 0 | 18 | 0 | 0 | 18 |

Article/Letter to the editor

Alves, RDAM, Demmers, J.A.A., Bezstarosti, K., Eerden, B.C.J. van der, Verhaar, J.A.N., Eijken, M & Leeuwen, J.P.T.M. van (2011). Unraveling the Human Bone Microenvironment beyond the Classical Extracellular Matrix Proteins: A Human Bone Protein Library. *Journal of Proteome Research*, 10(10), 4725-4733.

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EMC MGC-02-26-01 - Forensic molecular biology**Programme (brief description)**

The Department of Forensic Molecular Biology is a joint initiative of the Erasmus University Medical Center (Erasmus MC), the Erasmus University and the Netherlands Forensic Institute (NFI). We are using state-of-art technologies in genetics and genomics to answer questions in human biology that are of fundamental scientific interest and in addition provide potential applications to forensic sciences. Research topics are initiated by more current issues in forensic molecular biology such as the identification of the type of tissue and the age of a sample found at the crime scene, or the identification and interpretation of a male component using Y chromosome genetic information, or the identification of the geographic ancestry of an unknown DNA sample using genetic information, but also by more future issues such as the potential use of genetic information that indirectly or directly allows prediction of externally visible characteristics of humans. In addition, we study lethal disorders and unexplained death to understand their biology and to develop biomarkers for future molecular autopsy. We are also using human genetic variation to investigate relationships, origins and migration history of human populations and are additionally interested in footprints of local adaptation and natural selection in the human genome.

Key Figures

| Department | Sp1 | Sp2 | Sp3 | Sp4 | SpTot | T1 | T2 | T3 | Ttot | IS | FS | OP | Ptot |
|----------------------------|-------------|------------|------------|------------|--------------|-----------|-----------|-----------|-------------|-----------|-----------|-----------|-------------|
| forensic molecular biology | 6,75 | 2 | | | 8,75 | | | | | | | | |
| Total | 6,75 | 2 | | | 8,75 | 0 | 0 | 0 | 0 | 15 | 5 | 0 | 20 |

Article/Letter to the editor

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EMC MGC-02-52-01-A - Clinical and experimental aspects of embryogenesis and early placental development

Programme (brief description)

General objectives: To study the etiology of abnormal pregnancy outcome and gene-environmental interactions in relation to abnormal embryogenesis and first trimester malplacentation in particular. Acquired knowledge is used to develop programs of prevention of abnormal pregnancy outcome by risk selection and intervention before pregnancy (preconception) care and in early pregnancy (prenatal screening)

The following objectives are addressed:

a: the significance of 3D and 4D real-time colour Doppler ultrasound as well as the use of a Barco I-Space, a virtual reality system that allows binocular depth perception, for the study of normal and abnormal embryogenesis and placentation in early pregnancy (in collaboration with the department of Bioinformatics)

b: genetic and immunological aspects of abnormal placental development, subsequent suboptimal fetal growth and development and maternal pregnancy complications (in collaboration with depts. Clinical Genetics, Epidemiology and Generation R)

c: psychological and medico-ethical aspects of preconception care and prenatal medicine (in collaboration with the Depts of Bioethics and Medical Psychology)

d: feasibility of hospital and community programs of preconception care and prenatal screening

Key Figures

| Department | Sp1 | Sp2 | Sp3 | Sp4 | SpTot | T1 | T2 | T3 | Ttot | IS | FS | OP | Ptot |
|------------------------|-------------|-------------|------------|------------|--------------|-----------|-----------|-----------|-------------|-----------|-----------|-----------|-------------|
| gynaecology/obstetrics | 1,38 | 0,01 | | | 1,39 | | | | | | | | |
| Total | 1,38 | 0,01 | | | 1,39 | 7 | 0 | 0 | 7 | 73 | 7 | 3 | 90 |

Thesis

Bakker, R.. *Maternal lifestyle and pregnancy complication*. EUR. Prom./coprom.: Prof. Dr. E.A.P. Steegers.

Bliek, J.B. (2011, maart 02). *Folate related risk factors and orofacial clefting in human*. EUR (136 pag.). Prom./coprom.: Prof. Dr. E.A.P. Steegers, Prof. Dr. R.P.M. Steegers-Theunissen & Dr. J.E.M.M. de Klein.

Gaugler-Senden, I.P.M.. *Severe early onset preeclampsia*. EUR. Prom./coprom.: Prof. Dr. E.A.P. Steegers.

Hoedjes, M.. *Maternal Quality of Life, Lifestyle, and Interventions after Complicated Pregnancies*. EUR. Prom./coprom.: Prof. Dr. J.D.F. Habbema & Prof. Dr. E.A.P. Steegers.

Rousian, M.. *Embryonic Development in Virtual Reality*. EUR. Prom./coprom.: Prof. Dr. E.A.P. Steegers & Dr. N. Exalto.

Schoonen, H.M.H.J.D.. *Prenatal screening for Down syndrome and structural congenital anomalies in the Netherlands*. EUR. Prom./coprom.: Prof. Dr. H.J. de Koning & Prof. Dr. E.A.P. Steegers.

Smedts, H.P.M.. *Congenital heart disease, vascular risk factors and medication*. EUR. Prom./coprom.: Prof. Dr. R.P.M. Steegers-Theunissen & Prof. Dr. E.A.P. Steegers.

Article/Letter to the editor

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Part of book - abstract

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Report

Poeran, V.J.J., Birnie, E., Denktas, S., Steegers, E.A.P. & Bonsel, G.J. (2011). *Perinatale gezondheid in Rotterdam; nulmeting periode 2000-2007*. Rotterdam: Gemeente Rotterdam.

EMC MGC-02-53-01-A - Development disorders and congenital malformations

Programme (brief description)

The aim of this programme is to gain insight into the causes and effects of developmental disorders and congenital malformations. Various methods are employed. The first subprogramme (a) is based on a toxicologically-induced abnormal, or an existing and inbred phenotype, respectively, and attempts to map the genotype and associated gene products. Human material is included in the analyses. The second subprogramme (b) attempts to identify candidate genes, to analyse their products, and to define the process leading to the associated (abnormal) phenotype, using human material harvested during operative repair. Subprogrammes (c and d) assess the short-term and long-term medical, psychosocial and socially-relevant effects of the treatment of congenital malformations.

Subprogrammes:

Translational

a. The role of hormones and that of the genes expressed during normal and abnormal lung development are studied in a reproducible rat model of abnormal lung development induced by Nitrofen, in transgenic mice, and in organotypic cultures of embryonic lung buds.

Group leader: Dr. R. Rottier

b. To elucidate the phenotype-genotype relationship of derivatives of the foregut human material collected during operative repair, and combined with genomics and proteomics approaches to identify mutations. These data are combined with data from animal experiments using mutant mice evaluating the role of candidate genes during normal and abnormal phenotype expression.

Group leader: Dr. A. de Klein

Clinical studies

c1. Short-term studies: evidence based pharmacotherapy and care with a focus on pain and sedation in critically ill patients in the perioperative period.

Group leaders: Dr. M. van Dijk

Dr. S. de Wildt

c2. Long-term studies: somatic and psychosocial development; quality of life and parental support, using a unique prospective longitudinal database of over 1200 newborns with major congenital anomalies.

Group leader: Dr. H. Meijers-IJsselstijn

d. Surgical treatment and evaluation of congenital malformations.

New minimal access surgery and tissue engineering techniques are developed for the treatment of congenital malformations. These techniques are evaluated in randomized controlled trials.

Group leader: Prof. dr. R.M.H. Wijnen

Key Figures

| <i>Department</i> | <i>Sp1</i> | <i>Sp2</i> | <i>Sp3</i> | <i>Sp4</i> | <i>SpTot</i> | <i>T1</i> | <i>T2</i> | <i>T3</i> | <i>Ttot</i> | <i>IS</i> | <i>FS</i> | <i>OP</i> | <i>Ptot</i> |
|---------------------|--------------|-------------|------------|-------------|--------------|-----------|-----------|-----------|-------------|-----------|-----------|-----------|-------------|
| medical informatics | 0,13 | | | | 0,13 | | | | | | | | |
| pediatric surgery | 12,19 | 3,33 | | 6,71 | 22,23 | | | | | | | | |
| Total | 12,32 | 3,33 | | 6,71 | 22,36 | 7 | 1 | 0 | 8 | 41 | 4 | 2 | 55 |

Thesis

Boerlage, A.A. (2011, december 16). *Having a feel for others' pain*. EUR (146 pag.). Prom./coprom.: Prof. Dr. D. Tibboel & M. van Dijk.

Calis, E.A.C. (2011, oktober 05). *Correlates of lower respiratory tract infections and nutritional state in children with severe generalized cerebral palsy and intellectual disability*. EUR. Prom./coprom.: Prof. Dr. H.M. Evenhuis & Prof. Dr. D. Tibboel.

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Eijck, F.C. van. *Strategies and trends in the treatment of (giant) omphalocele*. EUR. Prom./coprom.: Prof.dr. R.M.H. Wijnen.

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Article/Letter to the editor

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EMC MGC-02-82-01 - Reproduction and development

Programme (brief description)

The research program is focused on gonadal development, gametogenesis and early embryogenesis in mammalian species. In doing so, the program is directly linked to clinical aspects of sex differentiation, male and female infertility, the early stages of human development, and stem cells. Hence, it involves collaboration with the clinical disciplines Pediatric Endocrinology, Gynecology and Obstetrics, Urology, Experimental Pathology, and Clinical Genetics. Members of the department participate in the Erasmus Stem Cell Institute and the Optical Imaging Centre

Gametogenesis (spermatogenesis and oogenesis) includes mitotic expansion of cells, meiotic recombination of genetic information, and unfolding of dedicated gene expression programs. Molecular and cellular factors implicated in control of gametogenesis are studied in genetically modified mouse models, also using cellular imaging technology. To facilitate transfer of knowledge to the clinic, the focus is on genes and proteins that are conserved between mouse and human. We study chromatin rearrangements and gene expression in gametogenesis and early embryogenesis, which is highly relevant in relation to an evaluation of possible risk factors associated with application of assisted reproduction techniques.

In mammalian female cells, one of two X chromosomes is inactivated, in relation to gene dosage compensation between female cells (XX) and male cells (XY). We are investigating the molecular mechanisms involved in this X chromosome inactivation. This research impacts on clinical aspects of X-linked diseases, and the development of stem cells. We also study the behavior of X and Y chromosomes in meiotic prophase.

Key Figures

| <i>Department</i> | <i>Sp1</i> | <i>Sp2</i> | <i>Sp3</i> | <i>Sp4</i> | <i>SpTot</i> | <i>T1</i> | <i>T2</i> | <i>T3</i> | <i>Ttot</i> | <i>IS</i> | <i>FS</i> | <i>OP</i> | <i>Ptot</i> |
|------------------------------|------------|------------|------------|------------|--------------|-----------|-----------|-----------|-------------|-----------|-----------|-----------|-------------|
| reproduction and development | 4,3 | 13 | | | 17,3 | | | | | | | | |
| Total | 4,3 | 13 | | | 17,3 | 0 | 0 | 0 | 0 | 12 | 2 | 0 | 14 |

Article/Letter to the editor

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EMC MGC-02-96-01 - Identification and characterisation of disease genes

Programme (brief description)

Identification and characterisation of disease genes will offer the opportunity for genetic counselling of couples with an enhanced genetic risk. In this project we will develop methods to study 'single gene disorders' as well as polygenetic / multifactorial disorders in order to diagnose a (genetic) defect in patients and carriers. For several disorders the genetic and cellular defect will be studied by isolating the responsible gene (positional cloning) followed by characterizing of the cellular defect or by characterizing and purifying the protein involved and subsequent isolation of the gene. We will study the gene defects and this will allow us to study the relation between the gene mutation and the cellular defects (genotype / phenotype relation). These methods have been used successfully to elucidate the etiology and pathogenesis of the fragile X syndrome, tuberous sclerosis and Parkinson disease. Also, there is a collaboration with Prof.Dr. D. Tibboel of the Paediatrics Surgery Department (EMC MGC-02-53-01-A) to study the genetic backgrounds of various congenital anomalies. Furthermore, together with the Department of Ophthalmology (EMC OR-01-60-01) we are looking for genes responsible for various congenital eye diseases.

At the same time we are studying lysosomal storage disorders and genetic factors involved in neurogenetic disorders (together with the department of Neurology MM-03-44-01) and Epidemiology (NIHES-01-64-01). Within families with hereditary hand malformations the genetic defect will be searched for via positional cloning of the responsible gene(s). After genetic counselling is has been shown that 50-90% of parents with a high genetic risk decide not to have more children. For most parents this is a difficult decision and offering the opportunity of prenatal diagnosis (followed eventually by a termination of the pregnancy) is of great relief for the parents. The second purpose of this project is to develop fast and reliable methods for prenatal diagnosis of inborn/hereditary disorders.

Key Figures

| <i>Department</i> | <i>Sp1</i> | <i>Sp2</i> | <i>Sp3</i> | <i>Sp4</i> | <i>SpTot</i> | <i>T1</i> | <i>T2</i> | <i>T3</i> | <i>Ttot</i> | <i>IS</i> | <i>FS</i> | <i>OP</i> | <i>Ptot</i> |
|-------------------|--------------|------------|------------|-------------|--------------|-----------|-----------|-----------|-------------|------------|-----------|-----------|-------------|
| clinical genetics | 13,37 | 3 | | 1,07 | 17,44 | | | | | | | | |
| Total | 13,37 | 3 | | 1,07 | 17,44 | 6 | 0 | 0 | 6 | 134 | 6 | 1 | 147 |

Thesis

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Article/Letter to the editor

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