Research Annual Report 2009

Erasmus MC
University Medical Center Rotterdam

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

Pathophysiology and treatment of chloride channel diseases

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

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

Programme design in brief

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, 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 NER (and transcription) deficiencies: such as 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. A second goal is to identify 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

Department	*SP1*	*SP2*	*SP3*	*SP4*	*SPtot*	*T1*	*T2*	*T3*	*Ttot*	*IS*	*FS*	*OP*	*Ptot*
genetics	9,86	13,59	12,29	11,6	47,34								
Total	9,86	13,59	12,29	11,6	47,34	3	0	0	3	47	0	2	52

Thesis

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Article/Letter to the editor

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EMC MGC-02-02-01 – Bioinformatics

Programme design in brief

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	4,69	0,07	0	1,07	5,83								
neurosciences	0,93	0	0	0	0,93								
Total	5,62	0,07	0	1,07	6,76	1	0	0	1	14	1	2	18

Thesis

Verwoerd-Dikkeboom, C.M. (2009, mei 08). Virtual embryoscopy. EUR. Prom./coprom.: Prof.Dr. E.A.P. Steegers.

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Part of book - abstract

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

Programme design in brief

Research is aimed at two main interests; the development of the hemapoietic system and the development of the brain including the neural crest. Study of the hemapoietic 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 rearrangment 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	11,84	12,81	6,37	7,81	38,83								
genetics	1,79	0	0	0,21	2								
surgery	0,5	0	0	0	0,5								
Total	14,13	12,81	6,37	8,02	41,33	8	0	0	8	25	3	2	38

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

Programme design in brief
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 (OR-01-54-02), Cardiology and Surgery (COEUR-09).

Key figures

Department	*SP1*	*SP2*	*SP3*	*SP4*	*SPtot*	*T1*	*T2*	*T3*	*Ttot*	*IS*	*FS*	*OP*	*Ptot*
cell biology	7,37	4,18	0,95	2,23	14,73								
Total	7,37	4,18	0,95	2,23	14,73	4	0	0	4	25	1	0	30

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

Programme design in brief

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. In particular, we focus 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. Our studies focus on three related topics: (1) The role of SWI/SNF-class ATP-dependent chromatin remodeling complexes in transcription regulation during development and disease. (2) Transcription control by protein (de)ubiquitylation. (3) Mechanism of gene silencing by Polycomb group proteins. For many of our studies, we use Drosophila as a model organism because it allows an integrated combination of biochemistry, proteomics and developmental genetics. In addition, we investigate the mechanism of tumor suppression by hSNF5, a core subunit of human SWI/SNF remodeling complexes. For these studies we use human tumor cell lines and mouse models. Moreover, we investigate the role of protein (de)ubiquitylation in human cancer. Finally, we are interested in how Polycomb group silencers and chromatin remodeling factors mediate epigenetic control of the INK/ARF tumor suppression locus and its effect on human disease and (stem) cell differentiation.

Key figures

Department	*SP1*	*SP2*	*SP3*	*SP4*	*SPtot*	*T1*	*T2*	*T3*	*Ttot*	*IS*	*FS*	*OP*	*Ptot*
biochemistry	4,57	5,69	2,43	0	12,69								
cell biology	0,5	0	0	0	0,5								
Total	5,07	5,69	2,43	0	13,19	2	0	0	2	4	1	0	7

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EMC MGC-02-21-02 - Pathophysiology and treatment of chloride channel diseases

Programme design in brief

The transport of anions across cellular membranes is crucial for a broad range of functions, including transport of salt and water across epithelia, the regulation of cell volume, ionic homeostasis of intracellular organells, exocytosis, pH regulation and the control of electrical excitability of muscle and nerves. This programme focuses on chloride channels involved in transepithelial ion transport (CFTR, bestrophins) and cell volume regulation (VRAC). Mutation or dysregulation of these chloride channels underlies a large spectrum of diseases including cystic fibrosis (CF), secretory diarrhea (e.g. cholera), pulmonary oedema, asthma and retinal degeneration. How mutations in the CFTR chloride channel affect the folding, processing, membrane recycling and gating of this protein and its regulation of other ion transporters such as sodium-hydrogen and chloride-bicarbonate exchangers is studied by biochemical and electrophysiological techniques at the level of cultured cells and CF mouse models. A major new research goal of the group is the in vivo rescue of mutant-CFTR function in CF mice by pharmacological approaches, and the ex vivo testing of promising candidate drugs by bioelectric assays in rectal biopsies and nasal mucosa from cystic fibrosis patients (carried out in collaboration with Drs. M. Sinaasappel and H. Tiddens from the Sophia Children¿s Hospital; see EMC MM-04-54-07). In addition, new approaches are explored to prevent excessive salt-and water loss in diarrheal diseases by interfering with transmembrane signaling by microbial enterotoxins or by the development of specific CFTR channel inhibitors.

Key figures

Department	*SP1*	*SP2*	*SP3*	*SP4*	*SPtot*	*T1*	*T2*	*T3*	*Ttot*	*IS*	*FS*	*OP*	*Ptot*
biochemistry	1,25	2,47	1,07	0	4,79								
Total	1,25	2,47	1,07	0	4,79	0	0	0	0	4	0	0	4

Article/Letter to the editor

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EMC MGC-02-26-01 - Forensic molecular biology

Programme design in brief

The Department of Forensic Molecular Biology is a joined 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	5,8	0	0	2	7,8								
Total	5,8	0	0	2	7,8	0	0	0	0	17	1	3	21

Article/Letter to the editor

Aulchenko, Y.S., Struchalin, M.V., Belonogova, NM, Axenovich, T.I., Weedon, MN, Hofman, A., Uitterlinden, A.G., Kayser, M., Oostra, B.A., Duijn, C.M. van, Janssens, A.C.J.W. & Borodin, PM (2009). Predicting human height by Victorian and genomic methods. European Journal of Human Genetics, 17(8), 1070-1075.

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

Programme design in brief

General objectives: To study the etiology of abnormal pregnancy outcome and gene-environmental interactions in relation to abnormal embyogenesis 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 fetalgrowth and development and maternal pregnancy complications (in collaboration with depts. Clinical Genetics, Epidemiology and Generation R)
c: psychological and medico-ethical aspects of preconption 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	2,6	0,85	0	0	3,45								
Total	2,6	0,85	0	0	3,45	3	0	1	4	57	9	7	77

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

Programme design in brief

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 two subprogrammes (a, b) are based on a toxicologically-induced abnormal, or an existing and inbred phenotype, respectively, and attempt to map the genotype and associated gene products. Human material was recently included in the analyses. The third subprogramme (c) attempts ¿ on the basis of the phenotype¿ 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. Subprogramme (d) assesses the long-term medical, psychosocial and socially-relevant effects of the treatment of congenital malformations.
Subprogrammes:
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.
b. Pathophysiologic and genetic characterization of the development of chronic renal insufficiency, and its possible prevention and treatment. Mainly the spontaneously hypertensive fawn-hooded (FHH) rat strain is used, which develops proteinuria and kidney damage at an early age and dies prematurely from end-stage renal failure. The susceptibility to develop renal damage is influenced by at least five genes, named Rf-1 to Rf-5. Main efforts are currently directed at pathophysiologic changes induced by the five Rf-loci and at the identification and functional characterization of Rf-1 on rat chromosome 1.
c. 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.
d. Short-term and long-term follow-up studies ¿ somatically and psychosocially (quality of life) ¿ of congenital malformations, especially related to parental support, nutritional assessment, pain management in pediatric surgical patients in the perioperative period, and management of undescended testes.

Key figures

Department	*SP1*	*SP2*	*SP3*	*SP4*	*SPtot*	*T1*	*T2*	*T3*	*Ttot*	*IS*	*FS*	*OP*	*Ptot*
anesthesiology	0,5	0	0	0	0,5								
pediatric surgery	7,62	0	0	4,87	12,49								
rehabilitation	0	0	0	0,06	0,06								
Total	8,12	0	0	4,93	13,05	2	0	0	2	40	9	3	54

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

Programme design in brief

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

Department	*SP1*	*SP2*	*SP3*	*SP4*	*SPtot*	*T1*	*T2*	*T3*	*Ttot*	*IS*	*FS*	*OP*	*Ptot*
reproduction and development	4,26	3,5	0	0	7,76								
Total	4,26	3,5	0	0	7,76	2	0	0	2	14	2	1	19

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

Programme design in brief

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.
At the same time we are studying lysosomal storage disorders and genetic factors involved in neurogenetic disorders (together with the department of Neurology (NEU440201) and Epidemiology (GZZ640101). Within families with hereditary hand malformations the genetic defect will be searched for via positional cloning of the responsible gene(s). In animal models the embryonal development of the limb will be studied to gain more insight into the function of the identified genes (together with the department of Plastic Surgery (HKG500101) and Anatomy (ANA100306).
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

Department	*SP1*	*SP2*	*SP3*	*SP4*	*SPtot*	*T1*	*T2*	*T3*	*Ttot*	*IS*	*FS*	*OP*	*Ptot*
clinical genetics	9,5	6,26	1,88	0,4	18,04								
genetics	0	0,6	0	0	0,6								
Total	9,5	6,86	1,88	0,4	18,64	6	0	0	6	151	12	0	169

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