



# Research Annual Report 2012



## 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)*

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

The centre 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 and congenital craniofacial malformations.

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 Centre. 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. This will pave the way for personalized healthcare to be implemented between 2015 and 2020.

### *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>
bioinformatics		0,6			0,6								
genetics	11,75	46,47	5,98	4,8	69								
<b>Total</b>	<b>11,75</b>	<b>47,07</b>	<b>5,98</b>	<b>4,8</b>	<b>69,6</b>	<b>4</b>	<b>0</b>	<b>1</b>	<b>5</b>	<b>31</b>	<b>1</b>	<b>0</b>	<b>37</b>

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Biernat, M. (2012, april 19). *Baculoviral and marsupial CPD photolyases: SNA repair proteins with a circadian clock function*. UW (113 pag.). Prom./coprom.: JM Vlak & Prof. Dr. G.T.J. van der Horst.

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### *Article/Letter to the editor*

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## EMC MGC-02-02-01 – Bioinformatics

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### *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>
bioinformatics	5,81	1,57		3,03	10,41								
<b>Total</b>	<b>5,81</b>	<b>1,57</b>	<b>0</b>	<b>3,03</b>	<b>10,41</b>	<b>0</b>	<b>0</b>	<b>0</b>	<b>0</b>	<b>15</b>	<b>1</b>	<b>0</b>	<b>16</b>

### *Article/Letter to the editor*

Beveren, N.J.M., Krab, L.C., Swagemakers, S.M.A., Buitendijk, G.H.S., Boot, E, Spek, P.J. van der, Elgersma, Y. & Amelvoort, TAMJ van (2012). Functional Gene-Expression Analysis Shows Involvement of Schizophrenia-Relevant Pathways in Patients with 22q11 Deletion Syndrome. *PLoS One*, 7(3).

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Santegoets, L.A.M., Baars, R. van, Terlou, A., Heijmans - Antonissen, C, Swagemakers, S.M.A., Spek, P.J. van der, Ewing-Graham, P.C., Beurden, M. van, Meijden, W.I. van der, Helmerhorst, T.J.M. & Blok, L.J. (2012). Different DNA damage and cell cycle checkpoint control in low- and high-risk human papillomavirus infections of the vulva. *International Journal of Cancer*, 130(12), 2874-2885.

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Zheng, P., Spek, P.J. van der, Dirven, C.M.F., Willemsen, R. & Kros, J.M. (2012). Sinus venosus defect (SVD) identified in zebrafish Glut1 morphants by video imaging. *International Journal of Cardiology*, 154(3), E60-E61.

## 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*

<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	0,92				0,92								
bioinformatics	0,8				0,8								
cell biology	20,39	27,99	2,58	3	53,96								
genetics	1				1								
<b>Total</b>	<b>23,11</b>	<b>27,99</b>	<b>2,58</b>	<b>3</b>	<b>56,68</b>	<b>6</b>	<b>2</b>	<b>0</b>	<b>8</b>	<b>20</b>	<b>2</b>	<b>0</b>	<b>30</b>

### *Thesis*

Aghajaniifah, A. (2012, november 14). *The role of ZBP-89 in globin regulation*. EUR (163 pag.). Prom./coprom.: Prof. Dr. F.G. Grosveld.

Berg, I.M. van den (2012, februari 22). *Epigenetic reprogramming during human oocyte maturation and early human development*. EUR (189 pag.). Prom./coprom.: Prof. Dr. J.S.E. Laven & Prof. Dr. F.G. Grosveld.

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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***

<b><i>Department</i></b>	<b><i>Sp1</i></b>	<b><i>Sp2</i></b>	<b><i>Sp3</i></b>	<b><i>Sp4</i></b>	<b><i>SpTot</i></b>	<b><i>T1</i></b>	<b><i>T2</i></b>	<b><i>T3</i></b>	<b><i>Ttot</i></b>	<b><i>IS</i></b>	<b><i>FS</i></b>	<b><i>OP</i></b>	<b><i>Ptot</i></b>
cell biology	6,34	7,07	2,08	1	16,49								
<b><i>Total</i></b>	<b><i>6,34</i></b>	<b><i>7,07</i></b>	<b><i>2,08</i></b>	<b><i>1</i></b>	<b><i>16,49</i></b>	<b><i>6</i></b>	<b><i>1</i></b>	<b><i>0</i></b>	<b><i>7</i></b>	<b><i>19</i></b>	<b><i>0</i></b>	<b><i>0</i></b>	<b><i>26</i></b>

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## 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	12,25	7,46			19,71								
cell biology	0,33				0,33								
<b>Total</b>	<b>12,58</b>	<b>7,46</b>	<b>0</b>	<b>0</b>	<b>20,04</b>	<b>2</b>	<b>0</b>	<b>0</b>	<b>2</b>	<b>21</b>	<b>2</b>	<b>0</b>	<b>25</b>

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### *Article/Letter to the editor*

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

### **Programme (brief description)**

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**

<b>Department</b>	<b>Sp1</b>	<b>Sp2</b>	<b>Sp3</b>	<b>Sp4</b>	<b>SpTot</b>	<b>T1</b>	<b>T2</b>	<b>T3</b>	<b>Ttot</b>	<b>IS</b>	<b>FS</b>	<b>OP</b>	<b>Ptot</b>
forensic molecular biology	7,92	3,67			11,59								
<b>Total</b>	<b>7,92</b>	<b>3,67</b>	<b>0</b>	<b>0</b>	<b>11,59</b>	<b>0</b>	<b>0</b>	<b>0</b>	<b>0</b>	<b>14</b>	<b>5</b>	<b>0</b>	<b>19</b>

### **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*

<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>
gynaecology/ obstetrics	2,05	0,01			2,06								
<b>Total</b>	<b>2,05</b>	<b>0,01</b>	<b>0</b>	<b>0</b>	<b>2,06</b>	<b>3</b>	<b>0</b>	<b>0</b>	<b>3</b>	<b>73</b>	<b>6</b>	<b>6</b>	<b>88</b>

### *Thesis*

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### *Article/Letter to the editor*

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#### **Part of book - Abstract**

Boer, K. & Steegers, E.A.P. (2012). Preconceptiezorg. In *Obstetrie en gynaecologie- de voortplanting van de mens* (pp. 207-231). Amsterdam: Reed Business.

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#### **Book Editorship**

Heineman, M.J., Evers, J.L.H., Massuger, L.F. & Steegers, E.A.P. (Eds.). (2012). *Obstetrie en Gynaecologie - de voortplanting van de mens* (7e editie). Amsterdam: Reed Business.

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**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***

<b><i>Department</i></b>	<b><i>Sp1</i></b>	<b><i>Sp2</i></b>	<b><i>Sp3</i></b>	<b><i>Sp4</i></b>	<b><i>SpTot</i></b>	<b><i>T1</i></b>	<b><i>T2</i></b>	<b><i>T3</i></b>	<b><i>Ttot</i></b>	<b><i>IS</i></b>	<b><i>FS</i></b>	<b><i>OP</i></b>	<b><i>Ptot</i></b>
medical informatics	0,13				0,13								
pediatric surgery	14,43	3,33		6,71	24,47								
<b><i>Total</i></b>	<b><i>14,56</i></b>	<b><i>3,33</i></b>	<b><i>0</i></b>	<b><i>6,71</i></b>	<b><i>24,6</i></b>	<b><i>11</i></b>	<b><i>0</i></b>	<b><i>1</i></b>	<b><i>12</i></b>	<b><i>65</i></b>	<b><i>4</i></b>	<b><i>0</i></b>	<b><i>81</i></b>

***Thesis***

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#### **Article/Letter to the editor**

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

### *Programme (brief description)*

Het onderzoek en onderwijs van de afdeling is gericht op epigenetica, geslachtschromosomen, de vorming van eicellen en zaadcellen (gametogenese, met een focus op de reductiedeling, de meiose), de ontwikkeling van het vroege embryo, en embryonale stamcellen.

De evolutie van de geslachtschromosomen bij zoogdieren heeft geleid tot het heterologe chromosomenpaar X en Y. De laatste jaren is er toenemende aandacht voor consequenties van genetische activiteit van de geslachtschromosomen, in relatie tot de biologie van vrouw en man en de gevoeligheid voor ziekten. Hierbij moet vooral worden gedacht aan regulatie en dysregulatie op het niveau van de epigenetica.

De epigenetica beschrijft erfelijke veranderingen in de expressie van het genoom, niet gebaseerd op mutatie van DNA, maar op methylering van DNA en modificaties van de chromatinestructuur. Juist op het gebied van deze epigenetische aspecten zijn er belangrijke vragen over effecten van omgevingsfactoren en in vitro manipulatie van embryo's op de ontwikkeling van mensen. Ons onderzoek betreft onder meer de epigenetische regulatie van gendosering in embryonale cellen, en de regulatie van chromatinestructuren tijdens de gametogenese in relatie tot de ontwikkeling van het vroege embryo.

Als een belangrijke activiteit binnen het Erasmus Stem Cell Institute, verrichten wij tevens onderzoek naar embryonale stamcellen (ES cellen) en geïnduceerde pluripotente stamcellen (iPS cellen). Dit stamcelonderzoek beoogt het produceren van ES en iPS cellen van muizen voor fundamenteel onderzoek, alsmede humane iPS cellen die kunnen worden gebruikt voor onderzoek gericht op patiënten met specifieke genetische aandoeningen.

Aan het geneeskunde bachelor-curriculum, de BSc Nanobiology en het MSc Molecular Medicine programma, alsmede graduate opleidingen elders in Nederland en Europa, levert de afdeling vele bijdragen. Bij dit onderwijs wordt gestreefd naar een duidelijke link met het onderzoek van de afdeling.

### *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	8,2	15			23,2								
<b>Total</b>	<b>8,2</b>	<b>15</b>	<b>0</b>	<b>0</b>	<b>23,2</b>	<b>3</b>	<b>1</b>	<b>0</b>	<b>4</b>	<b>11</b>	<b>0</b>	<b>1</b>	<b>16</b>

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Barakat, T.S. (2012, september 12). *Living with two X chromosomes: of mice and women*. EUR (386 pag.). Prom./coprom.: Prof. Dr. J.A. Grootegoed & Prof. Dr. J.H. Gribnau.

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#### **Part of book - Abstract**

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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. The programme recognizes two research corner stones: 1) neurogenetic disorders; 2) congenital anomalies. In both research lines 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 identification of the responsible gene (Next Generation Sequencing) followed by functional studies.

Functional studies include in vitro cellular models and in vivo animal models (mouse, zebrafish). We will study the gene defects and this will allow us to study the relation between the gene mutation and the cellular defects. These methods have been used successfully to elucidate the etiology and pathogenesis of the fragile X syndrome, FXTAS, tuberous sclerosis, Hirschsprung disease, Lynch Syndrome and Parkinson disease.

Also, there is a collaboration with Prof.dr. D. Tibboel and Prof.dr. R. Wijnen 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. In addition, we are studying lysosomal storage disorders and genetic factors involved in neurogenetic disorders (together with the department of Neurology (NEU440201) and Epidemiology (GZZ640101).

### *Key Figures*

<b>Department</b>	<b>Sp1</b>	<b>Sp2</b>	<b>Sp3</b>	<b>Sp4</b>	<b>SpTot</b>	<b>T1</b>	<b>T2</b>	<b>T3</b>	<b>Ttot</b>	<b>IS</b>	<b>FS</b>	<b>OP</b>	<b>Ptot</b>
clinical genetics	27,11	5,5	1,88		34,49								
<b>Total</b>	<b>27,11</b>	<b>5,5</b>	<b>1,88</b>	<b>0</b>	<b>34,49</b>	<b>7</b>	<b>0</b>	<b>0</b>	<b>7</b>	<b>147</b>	<b>9</b>	<b>0</b>	<b>163</b>

### *Thesis*

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**Article/Letter to the editor**

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