

## PUBLICATIONS DES ÉQUIPES DE RECHERCHE DE

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**KATHOLIEKE UNIVERISTEIT LEUVEN**  
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**Prof. dr. Claudia BAGNI**

DE RUBEIS S, PASCIUTO E, LI KW, FERNANDEZ E, DI MARINO D, BUZZI A, OSTROFF LE, ZWARTKUIJS F, KLANN E, KOMIYAMA NH, GRANT S, ACHSEL T, POSTHUMA D, SMIT AB and **BAGNI C**

**CYFIP1 co-ordinates mRNA translation and cytoskeleton remodeling to ensure proper dendritic spine formation.**

**Neuron**, Vol. 79, pp.1169-1182. **Impact Factor: 16.403.**

DI MARINO D, ACHSEL T, LACOUX C, FALCONI M and **BAGNI C**.

**Molecular Dynamics simulations show how the FMRP Ile304Asn mutation destabilizes the KH2 domain structure and affects its function.**

**Journal of Biomolecular Structure and Dynamics**, Mar 25, [Epub ahead of print].

**Impact Factor: 4.986**

FERNANDEZ E, RAJAN N and **BAGNI C**

**The FMRP regulon: from targets to disease convergence.**

**Frontiers in Neuroscience**, in press. **Impact Factor: to be determined.**

**Prof. dr. Danny HUYLEBROECK**

VAN DEN BERGHE V, STAPPERS E, VANDESANDE B, DIMIDSCHSTEIN J, KROES R, FRANCIS A, CONIDI A, LESAGE F, DRIES R, CAZZOLA S, BERX G, KESSARIS N, VANDERHAEGHEN P, VAN IJCKEN W, GROSVELD FG, GOOSSENS S, HAIGH JJ, FISHELL G, GOFFINET A, AERTS S, **HUYLEBROECK D\***, **SEUNTJENS E\***. (\* shared senior authors).

**Directed migration of cortical interneurons depends on the cell-autonomous action of Sip1.**

**Neuron**, Vol. 77, nr. 1, pp. 70-82. **Impact Factor: 15.982.**

MCKINSEY GL, LINDTNER S, TRZCINSKI B, VISEL A, PENNACCHIO L, **HUYLEBROECK D**, HIGASHI Y, RUBENSTEIN JL.

**Dlx1&2-dependent expression of Zfhx1b (Sip1, Zeb2) regulates the fate switch between cortical and striatal interneurons.**

**Neuron**, Vol. 77, nr. 1, pp. 83-98. **Impact Factor: 15.982.**

SEGKLIA A, **SEUNTJENS E**, ELKOURIS M, TSALAVOS S, STAPPERS E, MITSIADIS TA, **HUYLEBROECK D**. REMBOUTSIKA E, GRAF D.

**Bmp7 regulates the survival, proliferation, and neurogenic properties of neural progenitor cells during corticogenesis in the mouse.**

**PLoS One**, Vol. 7, nr. 3:e34088. **Impact Factor: 4.730.**

GÓMEZ-HERREROS F, ROMERO-GRANADOS R, ZENG Z, ALVAREZ-QUILÓN A, QUINTERO C, JU L, UMANS L, VERMEIRE L, HUYLEBROECK D, CALDECOTT KW\*, CORTÉS-LEDESMA F\*. (\* shared senior authors).

**TDP2-dependent non-homologous end-joining protects against topoisomerase II-induced DNA breaks and genome instability in cells and in vivo.**

***PLOS Genetics*, Vol. 9, nr. 3, e1003226. Impact Factor: 8.167.**

PRADIER B, JEUB M, MARKERT A, MAUER D, TOLKSDORF K, VAN DE PUTTE T, SEUNTJENS E, GAILUS-DURNER V, FUCHS H, HRABE DE ANGELIS M, HUYLEBROECK D, BECK H, ZIMMER A, RÁCZ I.

**Smad-interacting protein 1 affects acute and tonic, but not chronic pain.**

***European Journal of Pain*, Vol. 18, nr. 2, pp. 249-257. Impact Factor: 3.218.**

## Universiteit Antwerpen (UA)

### **Prof. dr. Marc CRUTS**

MORI K., WENG S-M., ARZBERGER T., MAY S., RENTZSCH K., KREMMER E., SCHMID B., KRETZSCHMAR H.A., CRUTS M., VAN BROEKHOVEN C., HAASS C. and EDBAUER D.:

**The *C9orf72* GGGGCC Repeat is Translated into Aggregating Dipeptide-Repeat Proteins in FTLD/ALS.**

***Science*, Vol. 339, pp.1335-1338 (2013). Impact Factor: 31.477.**

CRUTS M., GIJSELINCK I., VAN LANGENHOVE T., VAN DER ZEE J. and VAN BROEKHOVEN C.:

**Current insights into the *C9orf72* repeat expansion diseases of the FTLD/ALS spectrum.**

***Trends in Neurosciences*, Vol. 36, pp. 450-459 (2013). Impact Factor: 12.902.**

SCHMID B., HRUSCHA A., HOGL S., STRATHMANN J., STRECKER K., VAN DER ZEE J., TEUCKE M., EIMER S., HEGERMANN J., KITTELmann M., KREMMER E., CRUTS M., SOLCHENBERGER B., HASENKAMP L., VAN BEBBER F., VAN BROEKHOVEN C., EDBAUER D., LICHTENTHALER S.F. and HAASS C.:

**Loss of ALS-associated TDP-43 in zebrafish causes muscle degeneration, vascular dysfunction, and reduced motor neuron axon outgrowth.**

***PNAS*, Vol. 110, pp. 4986-4991 (2013). Impact Factor: 9.809.**

MORI K., LAMMICH S., MACKENZIE I.R., FORNÉ I., ZILOW S., KRETZSCHMAR H.A., EDBAUER D., JANSENS J., KLEINBERGER G., CRUTS M., HERMS J, NEUMANN M., VAN BROEKHOVEN C., ARZBERGER T. and HAASS C.:

**hnRNP A3 binds to GGGGCC repeats and is a constituent of p62 positive/TDP43-negative inclusions in the hippocampus of patients with *C9orf72* mutations.**

***Acta Neuropathologica*, Vol. 125, pp. 413-423 (2013). Impact Factor: 9.777.**

VAN LANGENHOVE T., VAN DER ZEE J., GIJSELINCK I., ENGELBORGH S., VANDENBERGHE R., VANDENBULCKE M., DE BLECKER J., SIEBEN A., VERSIJP J., IVANOIU A., DERYCK O., WILLEMS C., DILLEN L., PHILTJENS S., MAES G., BÄUMER V., VAN DEN BROECK M., MATTHEIJSENS M., PEETERS K., MARTIN J-J., MICHTOTTE A., SANTENS P., DE JONGHE P., CRAS P., DE DEYN P., **CRUTS M.** and VAN BROECKHOVEN C.:

**Distinct clinical characteristics of C9orf72 expansion carriers compared to GRN, MAPT and nonmutation carriers in a Flanders-Belgian FTLD cohort.**

JAMA Neurology, Vol. 70, pp.365-373 (2013). **Impact Factor: 7.008.**

DILLEN,L., VAN LANGENHOVE,T., ENGELBORGH S., VANDENBULCKE,M., SARAFOV,S., TOURNEV,I., MERLIN,C., CRAS,P., VANDENBERGHE,R., DE DEYN,P., JORDANOVA,A., **CRUTS,M.**, VAN BROECKHOVEN,C., VAN DER ZEE,J., BELNEU CONSORTIUM,:  
**Explorative genetic study of UBQLN2 and PFN1 in an extended Flanders-Belgian cohort of Frontotemporal Lobar Degeneration patients.**

Neurobiology of Aging, Vol. 34, pp.1711e1-5 (2013). **Impact Factor: 4.853.**

VAN DER ZEE J., GIJSELINCK I., DILLEN L., VAN LANGENHOVE T., THEUNS J., ENGELBORGH S., PHILTJENS S., VANDENBULCKE M., SLEEGERS K., SIEBEN A., BÄUMER V., MAES G., CORSMIT E., BORRONI B., PADOVANI A., ARCHETTI S., PERNECZKY R., DIEHL-SCHMID J., DE MENDONCA A., MILTENBERGER-MILTENYI G., PEREIRA S., PIMENTEL J., NACMIAS B., BAGNOLI S., SORBI S., GRAFF C., CHIANG H.-H., WESTERLUND M., SANCHEZ-VALLE R., LLADO A., GELPI E., SANTANA I., ROSARIO ALMEIDA M., SANTIAGO B., FRISONI G., ZANETTI O., BONVICINI C., SYNOFZIK M., MAETZLER W., MÜLLER VOM HAGEN J., SCHÖLS L., HENEKA M.T., JESSEN F., MATEJ R., PAROBKOVA E., KOVACS G.G., STRÖBEL T., SARAFOV S., TOURNEV I., JORDANOVA A., DANEK A., ARZBERGER T., FABRIZI G.-M., TESTI S., SALMON E., SANTENS P., MARTIN J-J., CRAS P., VANDENBERGHE R., DE DEYN P.P., **CRUTS M.**, and VAN BROECKHOVEN C., ON BEHALF OF THE EUROPEAN EARLY-ONSET DEMENTIA (EOD) CONSORTIUM:

**A Pan-European study of the C9orf72 repeat associated with FTLD: geographic prevalence, genomic instability and intermediate repeats.**

Human Mutation, Vol. 34, pp.363-373 (2013). **Impact Factor: 5.122.**

CUYVERS E., BETTENS K., PHILTJENS S., VAN LANGENHOVE T., GIJSELINCK I., VAN DER ZEE J., ENGELBORGH S., VANDENBULCKE M., VAN DONGEN J., GEERTS N., MAES G., MATTHEIJSENS M., PEETERS K., CRAS P., VANDENBERGHE R., DE DEYN P., VAN BROECKHOVEN C., **CRUTS M.**, and SLEEGERS K.:  
**Investigating the role of rare heterozygous TREM2 variants in Alzheimer's disease and frontotemporal dementia.**

Neurobiology of Aging, Epub (2013). **Impact Factor: 4.853**

MORI K., ARZBERGER T., GRÄSSER F.A., GIJSELINCK I., MAY S., RENTZSCH K., WENG S-M., SCHLUDI M.H., VAN DER ZEE J., **CRUTS M.**, VAN BROECKHOVEN C., KREMMER E., KRETZSCHMAR H., HAASS C., and EDBAUER D.:  
**Bidirectional transcripts of the expanded C9orf72 hexanucleotide repeat are translated into aggregating dipeptide repeat proteins.**

Acta Neuropathologica, Vol. 126, pp. 881-893 (2013). **Impact factor: 9.777.**

BANZHAF-STRATHMANN J., CLAUS R., MÜCKE O., RENTZSCH K., VAN DER ZEE J., ENGELBORGHHS S., DE DEYN P.P., CRUTS M., VAN BROECKHOVEN C., PLASS C. and EDBAUER D.:  
**Promoter DNA methylation regulates progranulin expression and is altered in FTLD.**  
*Acta Neuropathologica Communications*, Vol. 1, pp. 16 (2013). **Impact Factor: not known.**

**Prof. dr. Vincent TIMMERMAN en Prof. dr. Peter DE JONGHE**

VINCENT TIMMERMAN, VIRGINIA E. CLOWES and EVAN REID.  
**Overlapping molecular pathological themes link Charcot-Marie-Tooth neuropathies and hereditary spastic paraplegias.**  
*Experimental Neurology*, Vol. 246, pp. 14-25. **Impact Factor: 4.650.**

LEONARDO ALMEIDA-SOUZA, BOB ASSELBERGH, VICKY DE WINTER, SOFIE GOETHALS, VINCENT TIMMERMAN and SOPHIE JANSENS.  
**HSPB1 facilitates the formation of non-centrosomal microtubules.**  
*Plus One*, Vol. 8, nr. 6, e66541- pp. 1-13. **Impact Factor: 3.730.**

RICARDO E. MADRID, ANN LOFGREN, JONATHAN BAETS and VINCENT TIMMERMAN.  
**Biopsy in a patient with PMP22 exon 2 mutation recapitulates pathology of trembler-J Mouse.**  
*Neuromuscular Disorders*, Vol. 23, pp. 345-348. **Impact Factor: 3.460.**

INÈS MADEMAN, TINE DECONONCK, ARGRIOS DINOPoulos, THOMAS VOIT, ULRIKE SCHARA, KOENRAAD DEVRIENDT, BJÖRN MEJJERS, EVELYNE LERUT, PETER DE JONGHE and JONATHAN BAETS.  
**De novo INF2 mutations expand the genetic spectrum of hereditary neuropathy with glomerulopathy.**  
*Neurology*, Vol. 81, pp. 1-6. **Impact Factor: 8.310.**

ANNE HOLMGREN, DELPHINE BOUHY, VICKY DE WINTER, BOB ASSELBERGH, JEAN-PIERRE TIMMERMANS, JOY IROBI and VINCENT TIMMERMAN.  
**Charcot-Marie-Tooth causing HSPB1 mutations increase Cdk5-mediated phosphorylation of neurofilaments.**  
*Acta Neuropathology*, Vol. 126(1), pp. 93-108. **Impact Factor: 9.730.**

DELPHINE BOUHY and VINCENT TIMMERMAN.  
**Animal models and therapeutic prospects for Charcot-Marie-Tooth disease.**  
*Annals of Neurology*, Vol. 74, nr. 3, pp. 391-396. **Impact Factor: 11.190.**

KRISTIEN PEETERS, IVAN LITIVINENKO, BOB ASSELBERGH, LEONARDO ALMEIDA-SOUZA, TEODORA CHAMOVA, THOMAS GEUENS, ELKE YDENS, MAGDALENA ZIMORI, JOYY IROBI, ELS DE VRIENDT, VICKY DE WINTER, TINNE OOMS, VINCENT TIMMERMAN, IVAILO TOURNEV and ALBENA JORDANOVA.

**Molecular defects in the motor adaptor BICD2 cause proximal spinal muscular atrophy with autosoma-dominant inheritance.**  
*The American Journal of Human Genetics*, Vol. 92, pp. 955-964. **Impact Factor: 11.200.**

ENRICO LEIPOLD, LUTZ LIEBMANN, G. CHRISTOPH KORENKE, THERESA HEINRICH, SEBASTIAN GIEBELMANN, JONATHAN BAETS, MATTHIAS EBBINGHAUS, R. OLIVER GORAL, TOMMY STÖDBERG, J. CHRISTOPHER HENNINGS, MARKUS BERGMANN, JANINE ALTMÜLLER, HOLGER THIELE, ANDREA WETZEL, PETER NÜRNBERG, VINCENT TIMMERMAN, PETER DE JONGHE, ROBERT BLUM, HANS-GEORG SCHAIBLE, JOACHIM WEIS, STEFAN H. HEINEMANN, CHRISTIAN A. HÜBNER and INGO KURTH.  
**A de novo gain-of-function mutation in SCN11A causes loss of pain perception.**  
*Nature Genetics*, Vol. 45(11), pp. 1399-1404. **Impact factor: 34.520.**

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**Dr. Fadel TISSIR**

TATIN F, TADDEI A, WESTON A, FUCHS E, DEVENPORT D, **TISSIR F**, MAKINEN T.

**Planar cell polarity protein Celsr1 regulates endothelial adherens junctions and directed cell rearrangements during lymphatic valve morphogenesis.**

**Developmental Cell**, (2013), Vol. 26, pp. 31-44. **Impact factor: 13.020.**

VARDARAJAN B\*, VERGOTE D\*, **TISSIR F\***, LOGUE M, YANG J, DAUDE N, ANDO K, ROGAEVA E, LEE J, CHENG R, BRION JP, GHANI M, SHI B, BALDWIN CT, KAR S, MAYEUX R, FRASER P, GOFFINET AM, GEORGE-HYSLOP PS, FARRER LA, WESTAWAY D.

**Role of p73 in Alzheimer disease: lack of association in mouse models or in human cohorts.**

**Molecular Neurodegeneration** (2013), Vol. 8, pp. 1-14 \*: Equal contribution. **Impact factor : 5.290.**

**TISSIR F & GOFFINET AM.**

**Shaping the nervous system: Role of the planar cell polarity genes.**

**Nature Reviews Neuroscience** (2013), Vol. 14, pp.525-535. **Impact factor : 31.370.**

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**(VUB)**

**Prof dr. Ilse SMOLDERS en Prof. dr. Ann MASSIE**

VAN LIEFFERINGE J., MASSIE A., PORTELLI J., DI GIOVANNI G. and **SMOLDERS I.**

**Are vesicular neurotransmitter transporters potential treatment targets for temporal lobe epilepsy?**

**Front Cell Neurosci**.(2013), Vol. 7, pp. 139. Review. **Impact factor: 4.469.**

LEWERENZ J., HEWETT S.J., HUANG Y., LAMBROS M., GOUT P.W., KALIVAS P.W., **MASSIE A.**, **SMOLDERS I.**, METHNER A., PERGANDE M., SMITH S.B., GANAPATHY V. and MAHER P.

**The Cystine/Glutamate Antiporter System  $x_c^-$  in Health and Disease: From Molecular Mechanisms to Novel Therapeutic Opportunities. Review.**

**Antioxid Redox Signal**(2013), Vol. 18, pp. 522-555. **Impact factor: 7.189.**