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Forskningsinteresser

Our team at the Danish Epilepsy Centre, Filadelfia aim to improve the quality of life, and develop new personalized treatment options, for individuals with monogenic epilepsies.

Our research strives to unravel the underlying mechanisms of genetic epilepsies, to understand genotype phenotype correlations and to find new treatment options.

Our main research areas of interest are:
1) gene discovery in neurodevelopmental disorders and epilepsy;
2) electro-clinical characterization of genetic epilepsies;
3) functional characterization of genetic variants to understand their pathomechanisms;
4) to explore genotype-phenotype-pharmacoresponse correlations;
5) to improve existing or develop new personalized therapies for individuals with genetic epilepsies.

Kvalifikationer

Research Management Course, Copenhagen Business School
mar. 2017 → jun. 2017

PhD , Medical Genetics, Københavns Universitet
2004 → 2008

MSc, Biomedicine, Syddansk Universitet
1997 → 2003

Publikationer

Incidence of Aicardi-Goutières syndrome and KCNT1-related epilepsy in Denmark

Møller, R. S., Zhao, L., Shoaff, J. R., Duno, M., Andersen, B. N., Nguyen, V., Fang, T. C., Kupelian, V. & Thorén, R., dec. 2022, I: Molecular Genetics and Metabolism Reports. 33, 6 s., 100924.

Parental mosaicism in epilepsies due to alleged de novo variants

Møller, R. S., Liebmann, N., Larsen, L. H. G., Stiller, M., Hentschel, J., Kako, N., Abdin, D., Di Donato, N., Pal, D. K., Zacher, P., Syrbe, S., Dahl, H. A. & Lemke, J. R., 1. jun. 2019, I: Epilepsia. 60, 6, s. e63-e66

From next generation sequencing to targeted treatment of non-acquired epilepsies

Møller, R. S., Hammer, T. B., Rubboli, G., Lemke, J. R. & Johannesen, K. M., mar. 2019, I: Expert Review of Molecular Diagnostics. 19, 3, s. 217-228

Mutations in GABRB3: From febrile seizures to epileptic encephalopathies

Møller, R. S., Wuttke, T. V., Helbig, I., Marini, C., Johannesen, K. M., Brilstra, E. H., Vaher, U., Borggraefe, I., Talvik, I., Talvik, T., Kluger, G., Francois, L. L., Lesca, G., de Bellescize, J., Blichfeldt, S., Chatron, N., Holert, N., Jacobs, J., Swinkels, M. E. M. & Betzler, C. & 22 flere, Syrbe, S., Nikanorova, M., Myers, C. T., Larsen, L. H. G., Vejzovic, S., Pendziwiat, M., von Spiczak, S., Hopkins, S., Dubbs, H., Mang, Y., Mukhin, K., Holthausen, H., van Gassen, K. L., Dahl, H. A., Tommerup, N., Mefford, H. C., Rubboli, G., Guerrini, R., Lemke, J. R., Lerche, H., Muhle, H. & Maljevic, S., 31. jan. 2017, I: Neurology. 88, 5, s. 483-492

Gene Panel Testing in Epileptic Encephalopathies and Familial Epilepsies

Møller, R. S., Larsen, L. H. G., Johannesen, K. M., Talvik, I., Talvik, T., Vaher, U., Miranda, M. J., Farooq, M., Nielsen, J. E. K., Svendsen, L. L., Kjelgaard, D. B., Linnet, K. M., Hao, Q., Uldall, P., Frangu, M., Tommerup, N., Baig, S. M., Abdullah, U., Born, A. P. & Gellert, P. & 12 flere, Nikanorova, M., Olofsson, K., Jepsen, B., Marjanovic, D., Al-Zehhawi, L. I. K., Peñalva, S. J., Krag-Olsen, B., Brusgaard, K., Hjalgrim, H., Rubboli, G., Pal, D. K. & Dahl, H. A., 2016, I: Molecular Syndromology. 7, 4, s. 210-219

Germline and somatic mutations in the MTOR gene in focal cortical dysplasia and epilepsy
Møller, R. S., Weckhuysen, S., Chipaux, M., Marsan, E., Taly, V., Bebin, E. M., Hiatt, S. M., Prokop, J. W., Bowling, K. M., Mei, D., Conti, V., de la Grange, P., Ferrand-Sorbets, S., Dorfmüller, G., Lambrecq, V., Larsen, L. H. G., LeGuern, E., Guerrini, R., Rubboli, G. & Cooper, G. M. & 1 flere, Baulac, S., 2016, I: Neurology: Genetics. 2, 6, 13 s., e118.

Precision Medicine: *SCN8A* Encephalopathy Treated with Sodium Channel Blockers
Møller, R. S. & Johannesen, K. M., 2016, I: Neurotherapeutics. 13, 1, s. 190-191

Mutations in KCNT1 cause a spectrum of focal epilepsies
Møller, R. S., Heron, S. E., Larsen, L. H. G., Lim, C. X., Ricos, M. G., Bayly, M. A., van Kempen, M. J. A., Klinkenberg, S., Andrews, I., Kelley, K., Ronen, G. M., Callen, D., McMahon, J. M., Yendle, S. C., Carvill, G. L., Mefford, H. C., Nabbout, R., Poduri, A., Striano, P. & Baglietto, M. G. & 11 flere, Zara, F., Smith, N. J., Pridmore, C., Gardella, E., Nikanorova, M., Dahl, H. A., Gellert, P., Scheffer, I. E., Gunning, B., Kragh-Olsen, B. & Dibbens, L. M., sep. 2015, I: Epilepsia. 56, 9, s. e114-e120

The contribution of next generation sequencing to epilepsy genetics
Møller, R. S., Dahl, H. A. & Helbig, I., 2015, I: Expert Review of Molecular Diagnostics. 15, 12, s. 1531-1538

The Phenotypic Spectrum Associated with Gabrb3 Mutations: From Febrile Seizures to Severe Epileptic Encephalopathies
Moller, R. S., Nikanorova, M., Brilstra, E. H., Vaher, U., Talvik, I., Talvik, T., Kluger, G., Lemke, J. R., Myers, C., Larsen, L. H. G., Pendziwiat, M., Mang, Y., van Gassen, K. L., Dahl, H. A., Rubboli, G., Tommerup, N., Helbig, I., Mefford, H. C. & Muhle, H., 2015, I: Epilepsia. 56, S1, s. 19 1 s., 0052.

Genetic determinants of common epilepsies: a meta-analysis of genome-wide association studies
Møller, R. S. (Medlem af forfattergruppering) & International League Against Epilepsy Consortium on Complex Epilepsies, sep. 2014, I: The Lancet Neurology. 13, 9, s. 893-903

De novo mutations in synaptic transmission genes including *DNM1* cause epileptic encephalopathies
Møller, R. S. (Medlem af forfattergruppering), EuroEPINOMICS RES Consortium, Epilepsy Phenome/Genome Project & Epi4K Consortium, 2014, I: American Journal of Human Genetics. 95, 4, s. 360-370

X-linked congenital ptosis and associated intellectual disability, short stature, microcephaly, cleft palate, digital and genital abnormalities define novel Xq25q26 duplication syndrome
Moller, R. S., Jensen, L. R., Maas, S. M., Filmus, J., Capurro, M., Hansen, C., Marcelis, C. L. M., Ravn, K., Andrieux, J., Mathieu, M., Kirchhoff, M., Rodningen, O. K., de Leeuw, N., Yntema, H. G., Froyen, G., Vandewalle, J., Ballon, K., Klopocki, E., Joss, S. & Tolmie, J. & 11 flere, Knegt, A. C., Lund, A. M., Hjalgrim, H., Kuss, A. W., Tommerup, N., Ullmann, R., de Brouwer, A. P. M., Stromme, P., Kjaergaard, S., Tumer, Z. & Kleefstra, T., 2014, I: Human Genetics. 133, 5, s. 625-638

Exon-disrupting deletions of NRXN1 in idiopathic generalized epilepsy
Møller, R. S., YG, W., Klitten, L. L., H, T., H, M., WS, K., HC, M., A, F., M, K., P, W., D, D., S, S., IM, R., HE, W., JP, E., C, S., HJ, G., N, T., U, S. & H, L. & 3 flere, Hjalgrim, H., I, H. & T, S., feb. 2013, I: Epilepsia. 54, 2, s. 256-264 9 s.

Submikroskopiske kromosomforandringer disponerer til epilepsi
Møller, R. S. & Hjalgrim, H., 2011, I: Ugeskrift for Læger. 173, 16-17, s. 1201-4 4 s.

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Møller, R. S., Schneider, L. M., Hansen, C. P., Bugge, M., Ullmann, R., Tommerup, N. & Tümer, Z., jun. 2008, I: Epilepsia. 49, 6, s. 1091-1094

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Interstitial deletion of chromosome 4p associated with mild mental retardation, epilepsy and polymicrogyria of the left temporal lobe

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