Jens Michael Hertz
Professor, Chief Physician, MD, DMSc
Clinical Genetics, Odense University Hospital
Dep. of Clinical Research, Human Genetics
BRIDGE, Brain Research - Inter-Disciplinary Guided Excellence
Email: jens.michael.hertz@rsyd.dk
Mobile: 20277159
Phone: 65413191

Education and academic degrees

1983: MD, Aarhus University
1997: Specialist in clinical genetics
2009: DMSc., Aarhus University: ”Alport syndrome: Molecular genetic aspects”

Previous positions

1983-1984: Registrar, Terndrup and Hobro Hospital
1984-1995:MD, Institute of Human Genetics, Aarhus University
Nov. 1991:Consultant in clinical genetics for WHO in the Anhui-provins PR China
1995-1997:Senior registrar, Department of Clinical Genetics, Aarhus University Hospital
1997-2009:Consultant, Department of Clinical Genetics, Aarhus University Hospital
2006-2010:Associate professor, Institute of Human Genetics, Aarhus University
2009-2010:Head of Department, Department of Clinical Genetics, Aarhus University Hospital

Teaching

1985-2010:Human genetics, clinical genetics and statistics, Aarhus University
1995-2010:Appointed as external examiner for medical, dentist and biology students
1991-Teaching midwife students in genetics at University College Nordjylland
2010-Human and clinical genetics, University of Southern Denmark

Research evaluation

Evaluated 15 ph.d thesis and 20 master thesis.
Member of the evaluation committee for the research programme “Translational medicine” at the Faculty of Health Science, Aarhus University.
Chairman of the committee for 4 professor evaluations, 15 associate professor evaluations
Chairman for the evaluation committee for a doctor of honour evaluation, University of Southern Denmark.
Member of the evaluation committee for the Region of Southern Denmark ph.d. and post doc-scholarships.
Project evaluation for The Croatian Science Foundation, 2014.
Ad hoc review for 12 national and international scientific journals.

Administrative experience

1991-1994:Secretary for the Danish Society of Medical Genetics
2011-2012:Vice-chairman for the Danish Society of Medical Genetics
2012-2015:Chairman for the Danish Society of Medical Genetics
2012- Chairman for the Regional Committee on Health Research Ethics
2013- Danish representative the European Union of Medical Specialists (UEMS), Section for Clinical Genetics
2016-Member of the Odense University Hospital Research Counsil

Research education

Supervisor for 6 ph.d.-students (one ongoing), 5 master thesis students (one ongoing), and 11 medical students (two ongoing).

Publications

Frail inner limiting membrane maculopathy suggested to describe a new retinal Alport-like condition with two variants in three generations of females
Petersen, S. D., Belmouhand, M., Hertz, J. M., Fagerberg, C., Brasch-Andersen, C., Grauslund, J., Munier, F. L. & Larsen, M., 10. Jan 2024, (E-pub ahead of print) In: Ophthalmic Genetics.

Certain heterozygous variants in the kinase domain of the serine/threonine kinase NEK8 can cause an autosomal dominant form of polycystic kidney disease
Claus, L. R., Chen, C., Stallworth, J., Turner, J. L., Slaats, G., Hawks, A. L., Mabillard, H., Senum, S. R., Srikanth, S., Flanagan-Steet, H., Louie, R. J., Silver, J., Lerner-Ellis, J., Morel, C., Mighton, C., Sleutels, F., van Slegtenhorst, M., van Ham, T., Brooks, A. S. & Dorresteijn, E. M. & 28 others, Barakat, T. S., Dahan, K., Demoulin, N., Goffin, E. J., Olinger, E., Genomics England Research Consortium, Larsen, M., Hertz, J. M., Lilien, M. R., Obeidová, L., Seeman, T., Stone, H. K., Kerecuk, L., Gurgu, M., Yousef Yengej, F. A., Ammerlaan, C. M., Rookmaaker, M. B., Hanna, C., Rogers, R. C., Duran, K., Peters, E., Sayer, J. A., van Haaften, G., Harris, P. C., Ling, K., Mason, J. M., van Eerde, A. M. & Steet, R., Nov 2023, In: Kidney International. 104, 5, p. 995-1007

The clinical and molecular spectrum of the KDM6B-related neurodevelopmental disorder
Rots, D., Jakub, T. E., Keung, C., Jackson, A., Banka, S., Pfundt, R., de Vries, B. B. A., van Jaarsveld, R. H., Hopman, S. M. J., van Binsbergen, E., Valenzuela, I., Hempel, M., Bierhals, T., Kortüm, F., Lecoquierre, F., Goldenberg, A., Hertz, J. M., Andersen, C. B., Kibæk, M. & Prijoles, E. J. & 31 others, Stevenson, R. E., Everman, D. B., Patterson, W. G., Meng, L., Gijavanekar, C., De Dios, K., Lakhani, S., Levy, T., Wagner, M., Wieczorek, D., Benke, P. J., Lopez Garcia, M. S., Perrier, R., Sousa, S. B., Almeida, P. M., Simões, M. J., Isidor, B., Deb, W., Schmanski, A. A., Abdul-Rahman, O., Philippe, C., Bruel, A. L., Faivre, L., Vitobello, A., Thauvin, C., Smits, J. J., Garavelli, L., Caraffi, S. G., Peluso, F., Davis-Keppen, L. & Genomics England Research Consortium, 1. Jun 2023, In: American Journal of Human Genetics. 110, 6, p. 963-978

Whole exome sequencing of 28 families of Danish descent reveals novel candidate genes and pathways in developmental dysplasia of the hip
Dembic, M., van Brakel Andersen, L., Larsen, M. J., Mechlenburg, I., Søballe, K. & Hertz, J. M., Mar 2023, In: Molecular Genetics and Genomics. 298, 2, p. 329-342

Embracing Monogenic Parkinson's Disease: The MJFF Global Genetic PD Cohort
Vollstedt, E. J., Schaake, S., Lohmann, K., Padmanabhan, S., Brice, A., Lesage, S., Tesson, C., Vidailhet, M., Wurster, I., Hentati, F., Mirelman, A., Giladi, N., Marder, K., Waters, C., Fahn, S., Kasten, M., Brüggemann, N., Borsche, M., Foroud, T. & Tolosa, E. & 31 others, Garrido, A., Annesi, G., Gagliardi, M., Bozi, M., Stefanis, L., Ferreira, J. J., Correia Guedes, L., Avenali, M., Petrucci, S., Clark, L., Fedotova, E. Y., Abramycheva, N. Y., Alvarez, V., Menéndez-González, M., Jesús Maestre, S., Gómez-Garre, P., Mir, P., Belin, A. C., Ran, C., Lin, C. H., Kuo, M. C., Crosiers, D., Wszolek, Z. K., Ross, O. A., Jankovic, J., Nishioka, K., Funayama, M., Clarimon, J., Hertz, J. M., Petersen, M. S. & the MJFF Global Genetic Parkinson's Disease Study Group, Feb 2023, In: Movement Disorders. 38, 2, p. 286-303

Detection of DZIP1L mutations by whole-exome sequencing in consanguineous families with polycystic kidney disease
Hertz, J. M., Svenningsen, P., Dimke, H., Engelund, M. B., Nørgaard, H., Hansen, A., Marcussen, N., Thiesson, H. C., Bergmann, C. & Larsen, M. J., Nov 2022, In: Pediatric Nephrology. 37, 11, p. 2657-2665

A Novel AQP2 Sequence Variant Causing Aquaporin-2 Retention in the Cytoplasm and Autosomal Dominant Nephrogenic Diabetes Insipidus
Hinrichs, G. R., Baltzer, S., Pallien, T., Svenningsen, P., Dalgaard, E. B., Hertz, J. M., Bistrup, C., Jensen, B. L. & Klussmann, E., Oct 2022, In: Kidney International Reports. 7, 10, p. 2289-2294

Guidelines for Genetic Testing and Management of Alport Syndrome
Savige, J., Lipska-Zietkiewicz, B. S., Watson, E., Hertz, J. M., Deltas, C., Mari, F., Hilbert, P., Plevova, P., Byers, P., Cerkauskaite, A., Gregory, M., Cerkauskiene, R., Ljubanovic, D. G., Becherucci, F., Errichiello, C., Massella, L., Aiello, V., Lennon, R., Hopkinson, L. & Koziell, A. & 24 others, Lungu, A., Rothe, H. M., Hoefele, J., Zacchia, M., Martic, T. N., Gupta, A., van Eerde, A., Gear, S., Landini, S., Palazzo, V., Al-Rabadi, L., Claes, K., Corveleyn, A., Van Hoof, E., van Geel, M., Williams, M., Ashton, E., Belge, H., Ars, E., Bierzynska, A., Gangemi, C., Renieri, A., Storey, H. & Flinter, F., Jan 2022, In: Clinical Journal of the American Society of Nephrology. 17, 1, p. 143-154

Heterozygous Variants in Kinase Domain of NEK8 cause an Autosomal-Dominant Ciliopathy
Claus, L., Stallworth, J., van Jaarsveld, R., Turner, J., Hawks, A., May, M., Flanagan-Steet, H., Louie, R., Silver, J., Lerner-Ellise, J., Morel, C., Mighton, C., Ziegler, A., Barakat, S., Dahan, K., Demoulin, N., Jean Goffin, E., Larsen, M., Michael Hertz, J. & Lilien, M. & 14 others, Olinger, E., Sayer, J., Obeidová, L., Seeman, T., Senum, S., Hanna, C., Rogers, C., Duran, K., Peters, E., Harris, P., Mason, J., van Haaften, G., M. Van Eerde, A. & Steet, R., 2022, In: Nephrology Dialysis Transplantation. 37, Suppl. 3, 2 p., FC044.

HETEROZYGOUS VARIANTS IN KINASE DOMAIN OF NEK8 CAUSE AN AUTOSOMAL-DOMINANT CILIOPATHY
Claus, L. R., Stallworth, J., Van Jaarsveld, R. H., Louie, R., Silver, J., Lerner-Ellis, J., Morel, C., Mighton, C., Ziegler, A., Barakat, S., Dahan, K., Demoulin, N., Goffin, E., Larsen, M. J., Hertz, J. M., Lilien, M., Olinger, E., Sayer, J. A., Rogers, C. & Duran, K. J. & 4 others, Peters, E. D., Van Haaften, G., Steet, R. & Van Eerde, A. M., 1. Oct 2021, In: Pediatric Nephrology. 36, 10, p. 3299-3300 OP-36.

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Savige, J., Storey, H., Watson, E., Hertz, J. M., Deltas, C., Renieri, A., Mari, F., Hilbert, P., Plevova, P., Byers, P., Cerkauskaite, A., Gregory, M., Cerkauskiene, R., Ljubanovic, D. G., Becherucci, F., Errichiello, C., Massella, L., Aiello, V., Lennon, R. & Hopkinson, L. & 23 others, Koziell, A., Lungu, A., Rothe, H. M., Hoefele, J., Zacchia, M., Martic, T. N., Gupta, A., van Eerde, A., Gear, S., Landini, S., Palazzo, V., al-Rabadi, L., Claes, K., Corveleyn, A., Van Hoof, E., van Geel, M., Williams, M., Ashton, E., Belge, H., Ars, E., Bierzynska, A., Gangemi, C. & Lipska-Ziętkiewicz, B. S., Aug 2021, In: European Journal of Human Genetics. 29, 8, p. 1186-1197

Arvelige hårskaftsanomalier
Drivenes, J. L., Bygum, A., Hertz, J. M., Grimalt, R. & Puente de Pablo, N., 31. May 2021, In: Ugeskrift for Læger. 183, 20 , V02210125.

Skewness of X-chromosome inactivation increases with age and varies across birth cohorts in elderly Danish women
Mengel-From, J., Lindahl-Jacobsen, R., Nygaard, M., Sørensen, M., Ørstavik, K. H., Hertz, J. M., Andersen-Ranberg, K., Tan, Q. & Christensen, K., Feb 2021, In: Scientific Reports. 11, 9 p., 4326.

Biallelic variants in GLE1 with survival beyond neonatal period
Yates, T. M., Campeau, P. M., Ghoumid, J., Kibaek, M., Larsen, M. J., Smol, T., Albaba, S., Hertz, J. M. & Balasubramanian, M., Dec 2020, In: Clinical Genetics. 98, 6, p. 622-625

Low frequency of parental mosaicism in de novo COL4A5 mutations in X-linked Alport syndrome
Helle, O. M. B., Pedersen, T. H., Ousager, L. B., Thomassen, M. & Hertz, J. M., 1. Oct 2020, In: Molecular Genetics & Genomic Medicine. 8, 10, 7 p., e1452.

Expanding the cerebrovascular phenotype of the p.R258H variant in ACTA2 related hereditary thoracic aortic disease (HTAD)
Diness, B. R., Palmquist, R. N., Norling, R., Hove, H., Bundgaard, H., Hertz, J. M., Kondziella, D., Krieger, D., Dunø, M. & Grønborg, S., 15. Aug 2020, In: Journal of the Neurological Sciences. 415, 116897.

A non-pathogenic duplication of DMD exon 45-51, inserted in chromosome 17, in three Danish patients
Lauridsen, M. F., Koldby, K. M., Krogh, L. N., Graakjaer, J., Jensen, T. D., Fagerberg, C. & Hertz, J. M., 1. Jul 2019, In: European Journal of Human Genetics. 27, Supppl. 1, p. 326-327 2 p.

Familial multiple sclerosis patients have a shorter delay in diagnosis than sporadic cases
Steenhof, M., Stenager, E., Nielsen, N. M., Kyvik, K., Möller, S. & Hertz, J. M., 1. Jul 2019, In: Multiple Sclerosis and Related Disorders. 32, p. 97-102

Association between periacetabular osteotomy and hip dysplasia among relatives: a cross-sectional study
Simonsen, P. O., Hertz, J. M., Søballe, K. & Mechlenburg, I., Jul 2019, In: Hip International. 29, 4, p. 424-429

A novel PDGFRB sequence variant in a family with a mild form of primary familial brain calcification: A case report and a review of the literature
Mathorne, S. W., Sørensen, K., Fagerberg, C., Bode, M. & Hertz, J. M., 12. Apr 2019, In: BMC Neurology. 19, 6 p., 60.

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Genetic analysis of Charcot-Marie-Tooth disease in Denmark and the implementation of a next generation sequencing platform
Vaeth, S., Christensen, R., Dunø, M., Lildballe, D. L., Thorsen, K., Vissing, J., Svenstrup, K., Hertz, J. M., Andersen, H. & Jensen, U. B., Jan 2019, In: European Journal of Medical Genetics. 62, 1, p. 1-8

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Rasmussen, M., Lildballe, D. L., Brophy, P. D., Parida, M., Bonde, G., Hong, X., Clarke, J. C., Schneider, M., Sussman, C. R., Sunde, L., Hertz, J. M., Ramsing, M., Petersen, A., Cornell, R. A. & Manak, J. R., 2019, In: European Journal of Human Genetics. 26, p. 94-95 2 p., C18.6.

Anxiety and depression in Klinefelter syndrome: The impact of personality and social engagement
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DNA hypermethylation and differential gene expression associated with Klinefelter syndrome
Skakkebæk, A., Nielsen, M. M., Trolle, C., Vang, S., Hornshøj, H., Hedegaard, J., Wallentin, M., Bojesen, A., Hertz, J. M., Fedder, J., Østergaard, J. R., Pedersen, J. S. & Gravholt, C. H., 13. Sept 2018, In: Scientific Reports. 8, 15 p., 13740.

Properties and units in the clinical laboratory sciences part XXIV: Properties and units in clinical molecular genetics (technical report 2017)
Petersen, U. M., Padró-Miquel, A., Taylor, G., Hertz, J. M., Ceder, R., Fuentes-Arderiu, X. & den Dunnen, J. T., 1. Sept 2018, In: Clinica Chimica Acta. 484, p. 122-131

Compound heterozygous mutations in two different domains of ALDH18A1 do not affect the amino acid levels in a patient with hereditary spastic paraplegia
Steenhof, M., Kibæk, M., Larsen, M. J., Christensen, M., Lund, A. M., Brusgaard, K. & Hertz, J. M., Aug 2018, In: Neurogenetics. 19, 3, p. 145–149

Targeted Gene Sequencing and Whole-Exome Sequencing in Autopsied Fetuses with Prenatally Diagnosed Kidney Anomalies
Rasmussen, M., Sunde, L., Nielsen, M. L., Ramsing, M., Petersen, A., Hjortshøj, T. D., Olsen, T. E., Tabor, A., Hertz, J. M. , Johnsen, I., Sperling, L., Petersen, O. B., Jensen, U. B., Møller, F. G., Petersen, M. B. & Lildballe, D. L., Apr 2018, In: Clinical Genetics. 93, 4, p. 860-869

Hereditary spastic paraplegia type 8: Neuropathological findings
Pehrson, C., Hertz, J. M., Wirenfeldt, M., Stenager, E., Wermuth, L. & Winther Kristensen, B., Mar 2018, In: Brain Pathology. 28, 2, p. 292-294

Ichthyosis with Confetti Inherited from a Mosaic Father
Pallesen, K. A. U., Clemmensen, O., Fischer, J., Hertz, J. M. & Bygum, A., 12. Jan 2018, In: Acta Dermato-Venereologica. 98, 1, p. 130–131

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Gaist, D., Mogensen, J., Pedersen, E. G., Schrøder, H. D., Vissing, J., Andersen, H. & Hertz, J. M., 15. Aug 2017, In: Journal of the Neurological Sciences. 379, p. 217-218

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Kristensen, L. K., Larsen, L. A., Fagerberg, C., Hertz, J. M. & Christensen, K., 1. Jun 2017, In: Twin Research and Human Genetics. 20, 3, p. 216-225

Moebius sequence: a multidisciplinary clinical approach
Pedersen, L. K., Maimburg, R. D., Hertz, J. M., Gjørup, H., Pedersen, T. K., Møller-Madsen, B. & Østergaard, J. R., 6. Jan 2017, In: Orphanet Journal of Rare Diseases. 12, 11 p., 4.

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The first Danish family reported with an AQP5 mutation presenting diffuse non-epidermolytic palmoplantar keratoderma of Bothnian type, hyperhidrosis and frequent Corynebacterium infections: a case report
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Isobutyryl-CoA Dehydrogenase Deficiency Presenting with Significant Clinical Disease in Adulthood
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