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Klinisk Institut, forskningsenheden Human Genetik
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Uddannelse og akademiske grader

1983: Cand.med., Aarhus Universitet
1997: Speciallæge i klinisk genetik
2009: Dr.med., Aarhus Universitet: ”Alport syndrome: Molecular genetic aspects”

Tidligere ansættelser

1983-1984: Turnusuddannelse i Terndrup og Hobro
1984-1995: Læge ved Institut for Human Genetik, Aarhus Universitet
1991, nov.: Konsulent i klinisk genetik for WHO i Anhui-provinsen i Kina
1995-1997: Afdelingslæge, Klinisk Genetisk Afdeling, Århus Kommunehospital
1997-2009: Overlæge Klinisk Genetisk Afdeling, Århus Kommunehospital
2005-2009: Uddannelsesansvarlig overlæge, Klinisk Genetisk Afdeling, Århus Sygehus
2006-2010: Klinisk lektor i klinisk genetik, Det Sundhedsvidenskabelige Fakultet, AU
2009-2010: Ledende overlæge, Klinisk Genetisk Afdeling, Aarhus Universitetshospital

Undervisning

1985-2010:Undervisning i medicinsk genetik og statistik samt klinisk genetik, AU
1991-Undervisning i genetik ved Jordemoderuddannelsen, UCN
1995-2010:Beskikket som censor i medicinsk genetik ved lægeuddannelsen i Danmark
2006-2010:Beskikket som censor i medicinsk genetik ved tandlægeuddannelsen
2007-2010:Beskikket som censor ved Det Naturvidenskabelige Fakultet, Aarhus Universitet
2010-Undervisning og undervisningsadministration, Syddansk Universitet
2014-Beskikket som censor i genetik ved lægeuddannelsen i Danmark

Bedømmelse

Bedømmer på 15 ph.d.-afhandlinger og 20 master thesis afhandlinger.
Med i Styregruppen for Forskeruddannelsesprogrammet ”Translationel Medicin”, ved Det Sundhedsvidenskabelige Fakultet, Aarhus Universitet.
Formand for bedømmelsesudvalget ved 4 professorbedømmelser, og 15 lektorbedømmelser.
Formand for bedømmelsesudvalget ved æresdoktorbedømmelse og bedømmelse af adjungeret professor ved Syddansk Universitet.
Medlem af bedømmelsesudvalget for Region Syddanmarks ph.d. og post doc-pulje.
Projektbedømmelse for The Croatian Science Foundation, 2014.
Ad hoc review’er for 12 nationale og internationale tidsskrifter.

Bestyrelsesarbejde

1991-1994:Medlem af bestyrelsen og sekretær i Dansk Selskab for Medicinsk Genetik
2004-2012:Formand for DSMG's Uddannelsesudvalg og Hovedkursusleder
2011-2012:Næstformand for Dansk Selskab for Medicinsk Genetik
2012-2015Formand for Dansk Selskab for Medicinsk Genetik
2013- Formand for Den Videnskabsetiske Komité 2 i Region Syddanmark
2013- Dansk repræsentant i European Union of Medical Specialists (UEMS), Section of Clinical Genetics
2016-Medlem af OUH’s Forskningsråd.

Vejledning

Vejleder ved 5 afsluttede ph.d.-projekter, 8 kandidatspecialestuderende og 5 forskningsårsstuderende.
Aktuelt hovedvejleder for 1 ph.d.-studerende og 2 kandidatspecialestuderende.

Publikationer

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) I: 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 flereBarakat, 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, I: Kidney International. 104, 5, s. 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 flereStevenson, 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, I: American Journal of Human Genetics. 110, 6, s. 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, I: Molecular Genetics and Genomics. 298, 2, s. 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 flereGarrido, 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, I: Movement Disorders. 38, 2, s. 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, I: Pediatric Nephrology. 37, 11, s. 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., okt. 2022, I: Kidney International Reports. 7, 10, s. 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 flereLungu, 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, I: Clinical Journal of the American Society of Nephrology. 17, 1, s. 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 flereOlinger, 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, I: Nephrology Dialysis Transplantation. 37, Suppl. 3, 2 s., 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 flerePeters, E. D., Van Haaften, G., Steet, R. & Van Eerde, A. M., 1. okt. 2021, I: Pediatric Nephrology. 36, 10, s. 3299-3300 OP-36.

Consensus statement on standards and guidelines for the molecular diagnostics of Alport syndrome: refining the ACMG criteria
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 flereKoziell, 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, I: European Journal of Human Genetics. 29, 8, s. 1186-1197

Arvelige hårskaftsanomalier
Drivenes, J. L., Bygum, A., Hertz, J. M., Grimalt, R. & Puente de Pablo, N., 31. maj 2021, I: 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, I: Scientific Reports. 11, 9 s., 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, I: Clinical Genetics. 98, 6, s. 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. okt. 2020, I: Molecular Genetics & Genomic Medicine. 8, 10, 7 s., 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, I: 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, I: European Journal of Human Genetics. 27, Supppl. 1, s. 326-327 2 s.

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, I: Multiple Sclerosis and Related Disorders. 32, s. 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, I: Hip International. 29, 4, s. 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, I: BMC Neurology. 19, 6 s., 60.

Distribution of disease courses in familial vs sporadic multiple sclerosis
Steenhof, M., Nielsen, N. M., Stenager, E., Kyvik, K., Möller, S. & Hertz, J. M., mar. 2019, I: Acta Neurologica Scandinavica. 139, 3, s. 231-237

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, I: European Journal of Medical Genetics. 62, 1, s. 1-8

GREB1L and ROBO1 -Two novel genes associated with renal agenesis
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, I: European Journal of Human Genetics. 26, s. 94-95 2 s., C18.6.

Anxiety and depression in Klinefelter syndrome: The impact of personality and social engagement
Skakkebæk, A., Moore, P. J., Pedersen, A. D., Bojesen, A., Kristensen, M. K., Fedder, J., Hertz, J. M., Østergaard, J. R., Wallentin, M. & Gravholt, C. H., 1. nov. 2018, I: PLOS ONE. 13, 11, 11 s., e0206932.

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. sep. 2018, I: Scientific Reports. 8, 15 s., 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. sep. 2018, I: Clinica Chimica Acta. 484, s. 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, I: Neurogenetics. 19, 3, s. 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, I: Clinical Genetics. 93, 4, s. 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, I: Brain Pathology. 28, 2, s. 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, I: Acta Dermato-Venereologica. 98, 1, s. 130–131

Properties and units in the clinical laboratory sciences part XXIV. Properties and units in clinical molecular genetics (IUPAC Technical Report)
Petersen, U. M., Padró-Miquel, A., Taylor, G., Hertz, J. M., Ceder, R., Fuentes-Arderiu, X. & Den Dunnen, J. T., 2018, I: Pure and Applied Chemistry. 90, 7, s. 1199-1220

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

Chromosomal Aberrations in Monozygotic and Dizygotic Twins Versus Singletons in Denmark During 1968-2009
Kristensen, L. K., Larsen, L. A., Fagerberg, C., Hertz, J. M. & Christensen, K., 1. jun. 2017, I: Twin Research and Human Genetics. 20, 3, s. 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, I: Orphanet Journal of Rare Diseases. 12, 11 s., 4.

A Gene Implicated in Activation of Retinoic Acid Receptor Targets Is a Novel Renal Agenesis Gene in Humans
Brophy, P. D., Rasmussen, M., Parida, M., Bonde, G., Darbro, B. W., Hong, X., Clarke, J. C., Peterson, K. A., Denegre, J. , Schneider, M., Sussman, C. R., Sunde, L., Lildballe, D. L., Hertz, J. M., Cornell, R. A., Murray, S. A. & Manak, J. R., 1. jan. 2017, I: Genetics. 207, 1, s. 215-228

GREB1L and ROBO1 -Two Novel Genes Associated with Renal Agenesis
Rasmussen, M., Lildballe, D. L., Brophy, PD., Parida, M., Bonde, G., Hong, X., Clarke, JC., Schneider, M., Sussman, CR. , Sunde, L., Hertz, JM., Ramsing, M., Petersen, A., Cornell, RA. & Manak, JR., 2017.

Severe fluoropyrimidine toxicity due to novel and rare DPYD missense mutations, deletion and genomic amplification affecting DPD activity and mRNA splicing
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The association between gender and familial prevalence of hip dysplasia in Danish patients
El Jashi, R., Gustafson, M. B., Jakobsen, M. B., Lautrup, C. K., Hertz, J. M., Søballe, K. & Mechlenburg, I., 2017, I: Hip International. 27, 3, s. 299-304

The Danish HD Registry-a nationwide family registry of HD families in Denmark
Gilling, M., Budtz-Jørgensen, E., Boonen, S. E., Lildballe, D. L., Bojesen, A., Hertz, J. M. & Sørensen, S. A., 2017, I: Clinical Genetics. 92, 3, s. 338–341

The role of genes, intelligence, personality, and social engagement in cognitive performance in Klinefelter syndrome
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Epilepsy and cataplexy in Angelman syndrome: Genotype-phenotype correlations
Mertz, L. G. B., Christensen, R., Vogel, I., Hertz, J. M. & Ostergaard, J. R., sep. 2016, I: Research in Developmental Disabilities. 56, s. 177-182

X-Linked and Autosomal Recessive Alport Syndrome: Pathogenic Variant Features and Further Genotype-Phenotype Correlations
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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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Newborn with severe epidermolysis bullosa: to treat or not to treat?
Boesen, M. L., Bygum, A., Hertz, J. M. & Zachariassen, G., 26. apr. 2016, I: BMJ Case Reports. 2016, 4 s., 214727.

Odonto-onycho-dermal dysplasia in a patient homozygous for a WNT10A nonsense mutation and mild manifestations of ectodermal dysplasia in carriers of the mutation
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Fetal Kidney Anomalies: Next Generation Sequencing
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Fetal Kidney Anomalies: Next Generation Sequencing
Rasmussen, M., Sunde, L., Nielsen, M. L., Ramsing, M., Petersen, A., Hjortshøj, T. D., Olsen, T. E., Tabor, A., Hertz, J. MI., Johnsen, I., Sperling, L., Petersen, O. B., Jensen, U. B., Petersen, M. B. & Lidballe, D. L., 2016.

Genetic Analysis of 'PAX6-Negative' Individuals with Aniridia or Gillespie Syndrome
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