Jens Michael Hertz
Professor, specialeansvarlig overlæge, dr.med.
Klinisk Institut, forskningsenheden Human Genetik
Klinisk Genetisk Afdeling, Odense Universitetshospital
J. B. Winsløws Vej 4, 5000 Odense C
E-mail: jens.michael.hertz@rsyd.dk
Mobil: 20277159
Telefon: 65413191

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

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 Dermatovenereologica. 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

DOK7 congenital myasthenia may be associated with severe mitral valve insufficiency
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

Severe fluoropyrimidine toxicity due to novel and rare DPYD missense mutations, deletion and genomic amplification affecting DPD activity and mRNA splicing
van Kuilenburg, A. B. P., Meijer, J., Maurer, D., Dobritzsch, D., Meinsma, R., Los, M., Knegt, L. C., Zoetekouw, L., Jansen, R. L. H., Dezentjé, V., van Huis-Tanja, L. H., van Kampen, R. J. W., Hertz, J. M. & Hennekam, R. C. M., 2017, I : BBA Molecular Basis of Disease. 1863, 3, s. 721-730

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
Skakkebæk, A., Moore, P. J., Pedersen, A. D., Bojesen, A., Kristensen, M. K., Fedder, J., Laurberg, P., Hertz, J. M., Østergaard, J. R., Wallentin, M. & Gravholt, C. H., 2017, I : Brain and Behavior. 7, 3, 11 s., e00645.

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
Krøigård, A. B., Hetland, L. E., Clemmensen, O., Blaydon, D. C., Hertz, J. M. & Bygum, A., 3. jun. 2016, I : BMC Dermatology. 16, 6 s., 7.

Isobutyryl-CoA Dehydrogenase Deficiency Presenting with Significant Clinical Disease in Adulthood
Highland Nygaard, H., Gaist, D., Christensen, M., Dunø, M., Kjeldsen, M., Schrøder, H. D., Gregersen, N., Wibrand, F., Olsen, R. K. J. & Hertz, J. M., 5. maj 2016.

Corrigendum to "Neuroanatomical correlates of Klinefelter syndrome studied in relation to the neuropsychological profile" [NeuroImage:Clin 4 (2014) 1-9]
Skakkebæk, A., Gravholt, C. H., Rasmussen, P. M., Bojesen, A., Jensen, J. S., Fedder, J., Laurberg, P., Hertz, J. M., Østergaard, J. R., Pedersen, A. D. & Wallentin, M., 2016, I : NeuroImage: Clinical. 11, s. 52 1 s.

Epilepsy and cataplexy in Angelman syndrome: Genotype-phenotype correlations
Mertz, L. G. B., Christensen, R., Vogel, I., Hertz, J. M. & Ostergaard, J. R., 2016, I : Research in Developmental Disabilities. 56, s. 177-182

Erratum: Neuroanatomical correlates of Klinefelter syndrome studied in relation to the neuropsychological profile (NeuroImage:Clin(2014):4:1-9)
Skakkebæk, A., Gravholt, C. H., Rasmussen, P. M., Bojesen, A., Jensen, J. S., Fedder, J., Laurberg, P., Hertz, J. M., Ostergaard, J. R., Pedersen, A. D. & Wallentin, M., 2016, I : NeuroImage: Clinical. 11, s. 52 1 s.

Genetic Analysis of 'PAX6-Negative' Individuals with Aniridia or Gillespie Syndrome
Ansari, M., Rainger, J., Hanson, I. M., Williamson, K. A., Sharkey, F., Harewood, L., Sandilands, A., Clayton-Smith, J., Dollfus, H., Bitoun, P., Meire, F., Fantes, J., Franco, B., Lorenz, B., Taylor, D. S., Stewart, F., Willoughby, C. E., McEntagart, M., Khaw, P. T., Clericuzio, C. & 33 flere, Van Maldergem, L., Williams, D., Newbury-Ecob, R., Traboulsi, E. I., Silva, E. D., Madlom, M. M., Goudie, D. R., Fleck, B. W., Wieczorek, D., Kohlhase, J., McTrusty, A. D., Gardiner, C., Yale, C., Moore, A. T., Russell-Eggitt, I., Islam, L., Lees, M., Beales, P. L., Tuft, S. J., Solano, J. B., Splitt, M., Hertz, J. M., Prescott, T. E., Shears, D. J., Nischal, K. K., Doco-Fenzy, M., Prieur, F., Temple, I. K., Lachlan, K. L., Damante, G., Morrison, D. A., van Heyningen, V. & Fitzpatrick, D. R., 2016, I : PLOS ONE. 11, 4, 15 s.

Googlede lysfølsomhed og diagnosticerede sig selvmed erytropoietisk protoporfyri
Lindegaard Christiansen, A., Brusgaard, K., Hertz, J. M. & Bygum, A., 2016, I : Ugeskrift for Laeger. 177, 1A, V01150015.

Klinefelter syndrome has increased brain responses to auditory stimuli and motor output, but not to visual stimuli or Stroop adaptation
Wallentin, M., Skakkebæk, A., Bojesen, A., Fedder, J., Laurberg, P., Østergaard, J. R., Hertz, J. M., Pedersen, A. D. & Gravholt, C. H., 2016, I : NeuroImage: Clinical. 11, s. 239-251

Newborn with severe epidermolysis bullosa: to treat or not to treat?
Boesen, M. L., Bygum, A., Hertz, J. M. & Zachariassen, G., 2016, I : BMJ Case Reports. 2016, 4 s.

Odonto-onycho-dermal dysplasia in a patient homozygous for a WNT10A nonsense mutation and mild manifestations of ectodermal dysplasia in carriers of the mutation
Krøigård, A. B., Clemmensen, O., Gjørup, H., Hertz, J. M. & Bygum, A., 2016, I : BMC Dermatology. 16, 5 s., 3.

X-Linked and Autosomal Recessive Alport Syndrome: Pathogenic Variant Features and Further Genotype-Phenotype Correlations
Savige, J., Storey, H., Il Cheong, H., Gyung Kang, H., Park, E., Hilbert, P., Persikov, A., Torres-Fernandez, C., Ars, E., Torra, R., Hertz, J. M., Thomassen, M., Shagam, L., Wang, D., Wang, Y., Flinter, F. & Nagel, M., 2016, I : P L o S One. 11 , 9, 13 s., e0161802.

Noonans syndrom kan diagnosticere sklinisk og molekylærgenetisk
Krab Henningsen, M., Jelsig, A. M., Andersen, H., Brusgaard, K., Ousager, L. B. & Hertz, J. M., 3. aug. 2015, I : Ugeskrift for Laeger. 177, 32, s. V12140755

Noonans syndrom kan diagnosticeres klinisk og molekylærgenetisk
mk, H., Jelsig, A. M., Andersen, H., Brusgaard, K., Ousager, L. B. & Hertz, J. M., 3. aug. 2015, I : Ugeskrift for Læger. 177 , 24, s. 2-7 V12140755.

Anthropometry in Klinefelter syndrome - multifactorial influences due to CAG length, testosterone treatment and possibly intrauterine hypogonadism
Chang, S., Skakkebæk, A., Trolle, C., Bojesen, A., Hertz, J. M., Cohen, A., Hougaard, D. M., Wallentin, M., Pedersen, A. D., Ostergaard, J. R. & Gravholt, C. H. J., mar. 2015, I : Journal of Clinical Endocrinology and Metabolism. 100, 3, s. E508-E517

Clinical utility gene card for: Alport syndrome - update 2014
Hertz, J. M., Thomassen, M., Storey, H. & Flinter, F., 2015, I : European Journal of Human Genetics. 23, 9, 4 s.

A Retrospective Study of Clinical and Mutational Findings in 45 Danish Families with Ectodermal Dysplasia
Svendsen, M. T., Henningsen, E., Hertz, J. M., Vestergaard Grejsen, D. & Bygum, A., 4. feb. 2014, I : Acta Dermatovenereologica. 94, 5, s. 531-533

Association of CHRDL1 mutations and variants with X-linked megalocornea, Neuhäuser syndrome and central corneal thickness
Davidson, A. E., Cheong, S-S., Hysi, P. G., Venturini, C., Plagnol, V., Ruddle, J. B., Ali, H., Carnt, N., Gardner, J. C., Hassan, H., Gade, E., Kearns, L., Jelsig, A. M., Restori, M., Webb, T. R., Laws, D., Cosgrove, M., Hertz, J. M., Russell-Eggitt, I., Pilz, D. T. & 3 flere, Hammond, C. J., Tuft, S. J. & Hardcastle, A. J., 2014, I : PLOS ONE. 9, 8, s. e104163

Eating behavior, prenatal and postnatal growth in Angelman syndrome
Mertz, L. G. B., Christensen, R., Vogel, I., Hertz, J. M. & Ostergaard, J. R., 2014, I : Research in Developmental Disabilities. 35, 11, s. 2681-2690 10 s.

Epigenetic remodelling and dysregulation of DLGAP4 is linked with early-onset cerebellar ataxia
Minocherhomji, S., Hansen, C., Kim, H. G., Mang, Y., Bak, M., Guldberg, P., Papadopoulos, N., Eiberg, H., Doh, G. D., Mollgard, K., Hertz, J. M., Nielsen, J. E., Ropers, H. H., Tumer, Z., Tommerup, N., Kalscheuer, V. M. & Silahtaroglu, A., 2014, I : Human Molecular Genetics. 23, 23, s. 6163-6176

Manifestations of Gorlin-Goltz syndrome
Larsen, A. K., Mikkelsen, D., Hertz, J. M. & Bygum, A., 2014, I : Danish Medical Journal. 61, 5, s. A4829

Neuroanatomical correlates of Klinefelter syndrome studied in relation to the neuropsychological profile
Skakkebæk, A., Gravholt, C. H., Rasmussen, P. M., Bojesen, A., Jensen, J. S., Fedder, J., Laurberg, P. M., Hertz, J. M., Ostergaard, J. R., Pedersen, A. D. & Wallentin, M., 2014, I : NeuroImage. Clinical. 4, s. 1-9 9 s.

Neurodevelopmental outcome in Angelman syndrome: Genotype-phenotype correlations
Mertz, L. G. B., Thaulov, P., Trillingsgaard, A., Christensen, R., Vogel, I., Hertz, J. M. & Ostergaard, J. R., 2014, I : Research in Developmental Disabilities. 35, 7, s. 1742-1747 6 s.

Neuropsychology and brain morphology in Klinefelter syndrome - the impacts of genetics
Skakkebæk, A., Bojesen, A., Kristensen, M. K., Cohen, A., Hougaard, D. M., Hertz, J. M., Fedder, J., Laurberg, P., Wallentin, M., Østergaard, J. R., Pedersen, A. D. & Gravholt, C. H., 2014, I : Andrology. 2, 4, s. 632-640

Ny klassifikation og molekylærgenetisk viden om arvelig iktyose
Andersen, R. E., Hertz, J. M. & Bygum, A., 2014, I : Ugeskrift for Laeger. 176, 29, s. 2471-2474 4 s.

Angelman syndrome in Denmark. birth incidence, genetic findings, and age at diagnosis
Hertz, J. M., 2013, I : American Journal of Medical Genetics. Part A. 161, 9, s. 2197-2203 23913711.

Detailed investigations of proximal tubular function in Imerslund-Grasbeck syndrome
Storm, T., Zeitz, C., Cases, O., Amsellem, S., Verroust, P. J., Madsen, M. Ø., Benoist, J-F., Passemard, S., Lebon, S., Jønsson, I. M., Emma, F., Koldsø, H., Hertz, J. M., Nielsen, R., Christensen, E. & Kozyraki, R., 2013, I : B M C Medical Genetics. 14, 1, s. 1 11 s., 111.

Genomsekventering - klinisk anvendelse
Hertz, J. M., Gerdes, A-M., Grønskov, K., Thomassen, M. & Vogel, I., 2013, I : Ugeskrift for Laeger. 175, 12, s. 818

Muscle regeneration and inflammation in patients with facioscapulohumeral muscular dystrophy
Hauerslev, S., Ørngreen, M. C., Hertz, J. M., Vissing, J. & Krag, T. O., 2013, I : Acta Neurologica Scandinavica. 128, 3, s. 194-201 8 s.

Quaternary epitopes of α345(IV) collagen initiate Alport post-transplant anti-GBM nephritis
Olaru, F., Luo, W., Wang, X-P., Ge, L., Hertz, J. M., Kashtan, C. E., Sado, Y., Segal, Y., Hudson, B. G. & Borza, D-B., 2013, I : Journal of the American Society of Nephrology. 24, 6, s. 889-95

Renal phenotypic investigations of megalin-deficient patients: novel insights into tubular proteinuria and albumin filtration
Storm, T., Tranebjærg, L., Frykholm, C., Birn, H., Verroust, P. J., Nevéus, T., Sundelin, B., Hertz, J. M., Holmström, G., Ericson, K., Christensen, E. & Nielsen, R., 2013, I : Nephrology, Dialysis, Transplantation. 28, 3, s. 585-591

Best practice guidelines and recommendations on the molecular diagnosis of myotonic dystrophy types 1 and 2
Kamsteeg, E-J., Kress, W., Catalli, C., Hertz, J. M., Witsch-Baumgartner, M., Buckley, M. F., van Engelen, B. G. M., Schwartz, M. & Scheffer, H., 2012, I : European Journal of Human Genetics. 20, 12, s. 1203-8 6 s.

Clinical utility gene card for: Alport syndrome
Hertz, J. M., Thomassen, M., Storey, H. & Flinter, F., 2012, I : European Journal of Human Genetics. 20, 6

Endocrine function in 97 patients with myotonic dystrophy type 1
Ørngreen, M. C., Arlien-Søborg, P., Duno, M., Hertz, J. M. & Vissing, J., 2012, I : Journal of Neurology. 259, 5, s. 912-20 9 s.

Genetisk rådgivning
Hertz, J. M., Kjærgaard, S. & Jensen, P. K. A., 2012, *Medicinsk Genetik.* Nørby, S. & Jensen, P. K. A. (red.). 2. udg. København: FADL's Forlag, s. 163-171

Klinisk genetik
Skovby, F., Gerdes, A-M. & Hertz, J. M., 2012, *Basisbog i Diagnostiske Fag.* Hellebek, A. & Thomsen, H. S. (red.). 1. udg. København: Munksgaard , s. 167-196

Neurologiske og neuromuskulære sygdomme
Hertz, J. M. & Sørensen, S. A., 2012, *Medicinsk Genetik.* Nørby, S. & Jensen, P. K. A. (red.). 2. udg. København: FADL's Forlag, s. 285-306

A patient with cubilin deficiency
Storm, T., Emma, F., Verroust, P. J., Hertz, J. M., Nielsen, R. & Christensen, E. I., 6. jan. 2011, I : The New England Journal of Medicine. 364, 1, s. 89-91 3 s.

A founder synonymous COL7A1 mutation in three Danish families with dominant dystrophic epidermolysis bullosa pruriginosa identifies exonic regulatory sequences required for exon 87 splicing
Covaciu, C., Grosso, F., Pisaneschi, E., Zambruno, G., Gregersen, P. A., Sommerlund, M., Hertz, J. M. & Castiglia, D., 2011, I : British Journal of Dermatology. 165, 3, s. 678-682

Establishment of a pig fibroblast-derived cell line for locus-directed transgene expression in cell cultures and blastocysts
Jakobsen, J. E., Li, J., Moldt, B., Kragh, P. M., Callesen, H., Hertz, J. M., Bolund, L., Jørgensen, A. L., Mikkelsen, J. G. & Nielsen, A. L., 2011, I : Molecular Biology Reports. 38, 1, s. 151-61 11 s.

Genotype and phenotype in Klinefelter syndrome - impact of androgen receptor polymorphism and skewed X inactivation
Bojesen, A., Hertz, J. M. & Gravholt, C. H., 2011, I : International Journal of Andrology. Supplement. 34, 6 Pt 2, s. e642-8

Limited phenotypic variation of hypocalcified amelogenesis imperfecta in a Danish five-generation family with a novel FAM83H nonsense mutation
Haubek, D., Gjørup, H., Jensen, L. G., juncker, I., Nyegaard, M., Børglum, A., Poulsen, S. & Hertz, J. M., 2011, I : International Journal of Paediatric Dentistry (Online). 21, 6, s. 407-12 6 s.

Myoblasts generated by lentiviral mediated MyoD transduction of myotonic dystrophy type 1 (DM1) fibroblasts can be used for assays of therapeutic molecules
Larsen, J., Pettersson, O. J., Jakobsen, M., Thomsen, R., Pedersen, C. B., Hertz, J. M., Gregersen, N., Corydon, T. J. & Jensen, T. G., 2011, I : BMC Research Notes. 4, s. 490

Report from a new array-CGH centre: Including examples of interesting cases
Andersen, C. B., Hertz, J. M., Jelsig, A. M. & Fagerberg, C., 2011.

Skewed X-chromosome inactivation causing diagnostic misinterpretation in congenital nephrogenic diabetes insipidus
Faerch, M., Corydon, T. J., Rittig, S., Christensen, J. H., Hertz, J. M. & Jendle, J., 1. nov. 2010, I : Scandinavian Journal of Urology and Nephrology. 44, 5, s. 324-30 7 s.

Aarskog-Scott syndrome: clinical update and report of nine novel mutations of the FGD1 gene
Orrico, A., Galli, L., Faivre, L., Clayton-Smith, J., Azzarello-Burri, S. M., Hertz, J. M., Jacquemont, S., Taurisano, R., Arroyo Carrera, I., Tarantino, E., Devriendt, K., Melis, D., Thelle, T., Meinhardt, U. & Sorrentino, V., 1. feb. 2010, I : American Journal of Medical Genetics. Part A. 152A, 2, s. 313-8 6 s.

Hypocalcified type of amelogenesis imperfecta in a large family: clinical, radiographic, and histological findings, associated dento-facial anomalies, and resulting treatment load
Gjørup, H., Haubek, D., Hintze, H., Haukali, G., Løvschall, H., Hertz, J. M. & Poulsen, S., 18. maj 2009, I : Acta Odontologica Scandinavica. s. 1-8 8 s.

Pallister-Killian syndrome in a girl with mild developmental delay and mosaicism for hexasomy 12p
Vogel, I., Lyngbye, T. J. B., Nielsen, A., Pedersen, S. & Hertz, J. M., 1. mar. 2009, I : American Journal of Medical Genetics. Part A. 149A, 3, s. 510-4 5 s.

Alport syndrome. Molecular genetic aspects
Hertz, J. M., 2009, I : Danish Medical Bulletin (Online). 56, 3, s. 105-52 48 s.

Prenatal diagnostics in Arhus and Viborg counties after implementation of first trimester risk assessment.
Tørring, N., Jølving, L. R., Petersen, O. B. B., Holmskov, A., Hertz, J. M. & Uldbjerg, N., 7. jan. 2008, I : Ugeskrift for læger. 170, 1, s. 50-4 4 s., 170.

A novel missense mutation (G43S) in the switch I region of Rab27A causing Griscelli syndrome
Westbroek, W., Tuchman, M., Tinloy, B., De Wever, O., Vilboux, T., Hertz, J. M., Hasle, H., Heilmann, C., Helip-Wooley, A., Kleta, R. & Gahl, W. A., 2008, I : Molecular Genetics and Metabolism. 94, 2, s. 248-54 7 s.

Praenatal diagnostik i Arhus og Viborg Amter efter implementering af første trimester-risikovurdering
Tørring, N., Jølving, L. R., Petersen, O. B. B., Holmskov, A., Hertz, J. M. & Uldbjerg, N., 2008, I : Ugeskrift for Laeger. 170 , 1, s. 50-4 5 s.

X-linked hypohidrotic ectodermal dysplasia. Genetic and dental findings in 67 Danish patients from 19 families
Lexner, M. O., Bardow, A., juncker, I., Jensen, L. G., Almer, L. I., Kreiborg, S. & Hertz, J. M., 2008, I : The Application of Clinical Genetics. 74, 3, s. 252-9 8 s.

Anomalies of tooth formation in hypohidrotic ectodermal dysplasia
Lexner, M. O., Bardow, A., Hertz, J. M., Nielsen, L. & Kreiborg, S., 2007, I : International Journal of Paediatric Dentistry (Print). 17, 1, s. 10-8 9 s.

Anthropometric and cephalometric measurements in X-linked hypohidrotic ectodermal dysplasia
Lexner, M. O., Bardow, A., Bjorn-Jorgensen, J., Hertz, J. M., Almer, L. I. & Kreiborg, S., 2007, I : Orthodontics & Craniofacial Research. 10, 4, s. 203-15 13 s.

The variant inv(2)(p11.2q13) is a genuinely recurrent rearrangement but displays some breakpoint heterogeneity
Fickelscher, I., Liehr, T., Watts, K., Bryant, V., Barber, J. C. K., Heidemann, S., Siebert, R., Hertz, J. M., Tumer, Z. & Simon Thomas, N., 2007, I : American Journal of Human Genetics. 81, 4, s. 847-56 10 s.

Whole saliva in X-linked hypohidrotic ectodermal dysplasia
Lexner, M. O., Bardow, A., Hertz, J. M., Almer, L. I., Nauntofte, B. & Kreiborg, S., 2007, I : International Journal of Paediatric Dentistry (Print). 17, 3, s. 155-62 8 s.

Dopa-responsive dystonia and early-onset Parkinson's disease in a patient with GTP cyclohydrolase I deficiency?
Hjermind, L. E., Johannsen, L. G., Blau, N., Wevers, R. A., Lucking, C-B., Hertz, J. M., Friberg, L., Regeur, L., Nielsen, J. E. & Sørensen, S. A., 2006, I : Movement Disorders. 21, 5, s. 679-82 4 s.

Low frequency of Parkin, Tyrosine Hydroxylase, and GTP Cyclohydrolase I gene mutations in a Danish population of early-onset Parkinson's Disease
Hertz, J. M., Ostergaard, K., juncker, I., Pedersen, S., Romstad, A., Møller, L. B., Güttler, F. & Dupont, E., 2006, I : European Journal of Neurology. 13, 4, s. 385-90 6 s.

Alport syndrome caused by inversion of a 21 Mb fragment of the long arm of the X-chromosome comprising exon 9 through 51 of the COL4A5 gene
Hertz, J. M., Persson, U., juncker, I. & Segelmark, M., 2005, I : Human Genetics. 118, 1, s. 23-8 6 s.

Alport syndrome in southern Sweden
Persson, U., Hertz, J. M., Wieslander, J. & Segelmark, M., 2005, I : Clinical Nephrology. 64, 2, s. 85-90 6 s.

Cerebral parese--hvilken rolle spiller genetikken?
Rackauskaite, G., Balslev, T. & Hertz, J. M., 2005, I : Ugeskrift for Laeger. 167, 15, s. 1625-9 5 s.

Iron prophylaxis during pregnancy - How much iron is needed? A randomized dose-response study of 20-80 mg ferrous iron daily in pregnant women
Milman, N. T., Bergholt, T., Eriksen, L., Byg, K-E., Graudal, N., Pedersen, P. & Hertz, J., 2005, I : Acta Obstetricia et Gynecologica Scandinavica.

LGMD2I presenting with a characteristic Duchenne or Becker muscular dystrophy phenotype
Schwartz, M., Hertz, J. M., Sveen, M. L. & Vissing, J., 2005, I : Neurology. 64, 9, s. 1635-7 3 s.

Trisomy 13 due to rea(13q;13q) is caused by i(13) and not rob(13;13)(q10;q10) in the majority of cases
Bugge, M., deLozier-Blanchet, C., Bak, M., Brandt, C., Hertz, J. M., Nielsen, J. B., Duprez, L. & Petersen, M. B., 2005, I : American Journal of Medical Genetics. Part A. 132A, 3, s. 310-3 4 s.

Early onset, non-progressive, mild cerebellar ataxia co-segregating with a familial balanced translocation t(8;20)(p22;q13)
Hertz, J. M., Sivertsen, B. B., Silahtaroglu, A., Bugge, M., Kalscheuer, V., Weber, A., Wirth, J., Ropers, H-H., Tommerup, N. M. & Tümer, Z., 2004, I : Journal of Medical Genetics. 41, 3, s. e25

Patients with Goodpasture's disease have two normal COL4A3 alleles encoding the NC1 domain of the type IV collagen alpha 3 chain
Persson, U., Hertz, J. M., Carlsson, M., Hellmark, T., juncker, I., Wieslander, J. & Segelmark, M., 2004, I : Nephrology, Dialysis, Transplantation. 19, 8, s. 2030-5 6 s.

A novel mutation (R218Q) at the boundary between the N-terminal and the first transmembrane domain of the glycine receptor in a case of sporadic hyperekplexia
Miraglia Del Giudice, E., Coppola, G., Bellini, G., Ledaal, P., Hertz, J. M. & Pascotto, A., 2003, I : Journal of Medical Genetics. 40, 5, s. e71

X-linked Alport syndrome: natural history and genotype-phenotype correlations in girls and women belonging to 195 families: a "European Community Alport Syndrome Concerted Action" study
Jais, J. P., Knebelmann, B., Giatras, I., De Marchi, M., Rizzoni, G., Renieri, A., Weber, M., Gross, O., Netzer, K-O., Flinter, F., Pirson, Y., Dahan, K., Wieslander, J., Persson, U., Tryggvason, K., Martin, P., Hertz, J. M., Schröder, C., Sanak, M., Carvalho, M. F. & 4 flere, Saus, J., Antignac, C., Smeets, H. & Gubler, M. C., 2003, I : Journal of the American Society of Nephrology. 14, 10, s. 2603-10 8 s.

Pyknodysostose--faelles stamfar til en del af danske patienter: Udredning og molekylaergenetisk diagnostik
Haagerup, A., Christensen, M. F., Hertz, J. M. & Kruse, T. A., 2002, I : Ugeskrift for læger. 164, 7, s. 887-890

A new locus for Seckel syndrome on chromosome 18p11.31-q11.2
Børglum, A., Balslev, T., Haagerup, A., Birkebaek, N., Binderup, H. G., Kruse, T. A. & Hertz, J. M., 2001, I : European Journal of Human Genetics. 9, 10, s. 753-7 5 s.

Den molekylaergenetiske baggrund for en raekke arvelige kraniosynostoser og kondrodysplasier
Hertz, J. M., juncker, I., Christensen, L., Østergaard, J. R. & Jensen, P. K., 2001, I : Ugeskrift for Laeger. 163, 36, s. 4862-7 6 s.

Detection of mutations in the COL4A5 gene by SSCP in X-linked Alport syndrome
Hertz, J. M., Juncker, I., Persson, U., Matthijs, G., Schmidtke, J., Petersen, M. B., Kjeldsen, M. & Gregersen, N., 2001, I : Human Mutation. 18, 2, s. 141-8 8 s.

Meget svaer spinal muskelatrofi--type 0. En årsag til arthrogryposis multiplex congenita
Balslev, T., Hertz, J. M., Rackauskaite, G. & Sørensen, L. A., 2001, I : Ugeskrift for Laeger. 163, 41, s. 5679-80 2 s.

A novel splicing mutation in the V2 vasopressin receptor
Kamperis, K., Siggaard, C., Herlin, T., Nathan, E., Hertz, J. M. & Rittig, S., 2000, I : Pediatric Nephrology. 15, 1-2, s. 43-9 7 s.

Cathepsin K gene mutations and 1q21 haplotypes in at patients with pycnodysostosis in an outbred population
Haagerup, A., Hertz, J. M., Christensen, M. F., Binderup, H. G. & Kruse, T. A., 2000, I : European Journal of Human Genetics. 8, 6, s. 431-6 6 s.

X-linked Alport syndrome: natural history in 195 families and genotype- phenotype correlations in males
Jais, J. P., Knebelmann, B., Giatras, I., De Marchi, M., Rizzoni, G., Renieri, A., Weber, M., Gross, O., Netzer, K. O., Flinter, F., Pirson, Y., Verellen, C., Wieslander, J., Persson, U., Tryggvason, K., Martin, P., Hertz, J. M., Schröder, C., Sanak, M., Krejcova, S. & 5 flere, Carvalho, M. F., Saus, J., Antignac, C., Smeets, H. & Gubler, M. C., 2000, I : Journal of the American Society of Nephrology. 11, 4, s. 649-57 9 s.

Hereditaer neuropati med tendens til trykpareser
Smith, T. A., Rasmussen, K. & Hertz, J. M., 1999, I : Ugeskrift for Laeger. 161, 23, s. 3463-5 3 s.

A novel missense mutation (402C-->T) in exon 1 in the EDA gene in a family with X-linked hypohidrotic ectodermal dysplasia
Hertz, J. M., Nørgaard Hansen, K., Juncker, I., Kjeldsen, M. & Gregersen, N., 1998, I : Clinical Genetics. 53, 3, s. 205-9 5 s.

Apolipoprotein E alleles in mothers of trisomy 18 conceptuses
Hansen, C., Bugge, M., Brandt, C. A., Hertz, J. M., Tranebjaerg, L., Mikkelsen, M. & Petersen, M. B., 1998, I : Clinical Genetics. 53, 4, s. 321-2 2 s.

High mutation detection rate in the COL4A5 collagen gene in suspected Alport syndrome using PCR and direct DNA sequencing
Martin, P., Heiskari, N., Zhou, J., Leinonen, A., Tumelius, T., Hertz, J. M., Barker, D., Gregory, M., Atkin, C., Styrkarsdottir, U., Neumann, H., Springate, J., Shows, T., Pettersson, E. & Tryggvason, K., 1998, I : Journal of the American Society of Nephrology. 9, 12, s. 2291-301 11 s.

Juvenil neuronal ceroid lipofuscinosis
Ostergaard, J. R. & Hertz, J. M., 1998, I : Ugeskrift for Laeger. 160, 26, s. 3895-900 6 s.

Non-disjunction of chromosome 18
Bugge, M., Collins, A., Petersen, M. B., Fisher, J., Brandt, C., Hertz, J. M., Tranebjaerg, L., de Lozier-Blanchet, C., Nicolaides, P., Brøndum-Nielsen, K., Morton, N. & Mikkelsen, M., 1998, I : Human Molecular Genetics. 7, 4, s. 661-9 9 s.

Origin of nondisjunction in trisomy 8 and trisomy 8 mosaicism
Karadima, G., Bugge, M., Nicolaidis, P., Vassilopoulos, D., Avramopoulos, D., Grigoriadou, M., Albrecht, B., Passarge, E., Annerén, G., Blennow, E., Clausen, N., Galla-Voumvouraki, A., Tsezou, A., Kitsiou-Tzeli, S., Hahnemann, J. M., Hertz, J. M., Houge, G., Kuklík, M., Macek, M., Lacombe, D. & 5 flere, Miller, K., Moncla, A., López Pajares, I., Patsalis, P. C. & Petersen, M. B., 1998, I : European Journal of Human Genetics. 6, 5, s. 432-8 7 s.

Mutation pattern in the Bruton's tyrosine kinase gene in 26 unrelated patients with X-linked agammaglobulinemia
Vorechovský, I., Luo, L., Hertz, J. M., Frøland, S. S., Klemola, T., Fiorini, M., Quinti, I., Paganelli, R., Ozsahin, H., Hammarström, L., Webster, A. D. & Smith, C. I., 1997, I : Human Mutation. 9, 5, s. 418-25 8 s.

Quantification, by solid-phase minisequencing, of the telomeric and centromeric copies of the survival motor neuron gene in families with spinal muscular atrophy
Schwartz, M., Sørensen, N., Hansen, F. J., Hertz, J. M., Nørby, S., Tranebjaerg, L. & Skovby, F., 1997, I : Human Molecular Genetics. 6, 1, s. 99-104 6 s.

A nonsense mutation in the COL4A5 collagen gene in a family with X-linked juvenile Alport syndrome
Hertz, J. M., Heiskari, N., Zhou, J., Jensen, U. B. & Tryggvason, K., 1995, I : Kidney International. 47, 1, s. 327-32 6 s.

A TaqI and a BamHI polymorphism in the COL4A4 gene on chromosome 2q35-37
Hertz, J. M., Mariyama, M., Thomsen, A., Reeders, S. T. & Kruse, T. A., 1995, I : Clinical Genetics. 48, 3, s. 162-3 2 s.

Partial deletion 11q: report of a case with a large terminal deletion 11q21-qter without loss of telomeric sequences, and review of the literature
Hertz, J. M., Tommerup, N., Sørensen, F. B., Henriques, U. V., Nielsen, A. & Therkelsen, A. J., 1995, I : Clinical Genetics. 47, 5, s. 231-5 5 s.

Charcot-Marie-Tooth disease type 1A: the parental origin of a de novo 17p11.2-p12 duplication
Hertz, J. M., Børglum, A. D., Brandt, C. A., Flint, T. & Bisgaard, C., 1994, I : Clinical Genetics. 46, 4, s. 291-4 4 s.

The EUROGEM map of human chromosome 2
Flint, T. J., Hertz, J. M., Vergnaud, G., Orrù, S., Harvey, C. B., Bakker, B. & Kruse, T. A., 1994, I : European Journal of Human Genetics. 2, 3, s. 206-7 2 s.

Genetic analysis of repeated, biparental, diploid, hydatidiform moles
Sunde, L., Vejerslev, L. O., Jensen, M. P., Pedersen, S., Hertz, J. M. & Bolund, L., 1993, I : Cancer Genetics and Cytogenetics. 66, 1, s. 16-22 7 s.

Mutations in the codon for a conserved arginine-1563 in the COL4A5 collagen gene in Alport syndrome
Zhou, J., Gregory, M. C., Hertz, J. M., Barker, D. F., Atkin, C., Spencer, E. S. & Tryggvason, K., 1993, I : Kidney International. 43, 3, s. 722-9 8 s.

Complete amino acid sequence of the human alpha 5 (IV) collagen chain and identification of a single-base mutation in exon 23 converting glycine 521 in the collagenous domain to cysteine in an Alport syndrome patient
Zhou, J., Hertz, J. M., Leinonen, A. & Tryggvason, K., 1992, I : Journal of Biological Chemistry. 267, 18, s. 12475-81 7 s.

Mechanisms of ring chromosome formation in 11 cases of human ring chromosome 21
McGinniss, M. J., Kazazian, H. H., Stetten, G., Petersen, M. B., Boman, H., Engel, E., Greenberg, F., Hertz, J. M., Johnson, A. & Laca, Z., 1992, I : American Journal of Human Genetics. 50, 1, s. 15-28 14 s.

Mutation in the alpha 5(IV) collagen chain in juvenile-onset Alport syndrome without hearing loss or ocular lesions: detection by denaturing gradient gel electrophoresis of a PCR product
Zhou, J., Hertz, J. M. & Tryggvason, K., 1992, I : American Journal of Human Genetics. 50, 6, s. 1291-300 10 s.

Pericentric inversion of chromosome 12; a three family study
Haagerup, A. & Hertz, J. M., 1992, I : Human Genetics. 89, 3, s. 292-4 3 s.

Ring chromosome 13: lack of distinct syndromes based on different breakpoints on 13q
Brandt, C. A., Hertz, J. M., Petersen, M. B., Vogel, F., Noer, H. & Mikkelsen, M., 1992, I : Journal of Medical Genetics. 29, 10, s. 704-8 5 s.

Multipoint linkage analysis in X-linked Alport syndrome
Hertz, J. M., Kruse, T. A., Thomsen, A. & Spencer, E. S., 1991, I : Human Genetics. 88, 2, s. 157-61 5 s.

In situ hybridization analysis of isodicentric X-chromosomes with short arm fusion
Koch, J. E., Kølvraa, S., Hertz, J. M., Rasmussen, K., Gregersen, N., Fly, G. F. & Bolund, L. A., 1990, I : Clinical Genetics. 37, 6, s. 450-5 6 s.

Transabdominal chorion villus biopsi ved abnormt ultralydfund i 2. trimester
Hertz, J. M., Jensen, P. K., Henriques, U. & Maigaard, S., 1990, I : Ugeskrift for Laeger. 152, 1, s. 28-30 3 s.

Interstitial deletion of the short arm of chromosome 3. Fetal pathology and exclusion of the gene for beta-galactosidase-1 (GLB-1) from 3(p11----p14.2)
Hertz, J. M., Coerdt, W., Hahnemann, N. & Schwartz, M., 1988, I : Human Genetics. 79, 4, s. 389-91 3 s.

Prenatal cytogenetic diagnosis after transabdominal chorionic villus sampling in the first trimester
Therkelsen, A. J., Jensen, P. K., Hertz, J. M., Smidt-Jensen, S. & Hahnemann, N., 1988, I : Prenatal Diagnosis. 8, 1, s. 19-31 13 s.

Convoluted cells as a marker for maternal cell contamination in CVS cultures
Hertz, J. M., Jensen, P. K. & Therkelsen, A. J., 1987, I : Clinical Genetics. 31, 6, s. 410-2 3 s.

Familial transmission of a ring chromosome 21
Hertz, J. M., 1987, I : Clinical Genetics. 32, 1, s. 35-9 5 s.

Praenatal cytogenetisk diagnostik i første trimester
Jensen, P. K., Hertz, J. M., Hahnemann, N., Smidt-Jensen, S. & Therkelsen, A. J., 1987, I : Ugeskrift for Laeger. 149, 11, s. 718-21 4 s.

Admixture of maternal metaphases in first trimester direct chromosome preparations?
Jensen, P. K., Hertz, J. M. & Therkelsen, A. J., 1986, I : Prenatal Diagnosis. 6, 5, s. 383-5 3 s.

Interstitial deletion 1p as a result of a de novo reciprocal 1p;2p translocation
Hertz, J. M. & Jensen, P. H., 1985, I : European Journal of Medical Genetics. 28, 4, s. 228-30 3 s.