Main research areas

Craniofacial disorders, including on-going research field of the aspects of facial and cranial asymmetry.
Skeletal Dysplasias, In-born connective tissue defects; Neurofibromatosis von Recklinghausen (participation with Danish Cancer Society Research Center)

Employment

Chief Consultant

Department of Paediatrics and Adolescent Medicine

The Capital Region of Denmark

1 Jan 2017 → present

Research output

Neurocognitive functioning in adults with neurofibromatosis type 1- a nationwide population-based study
Doser, K., Jepsen, J. R. M., Kenborg, L., Miskowiak, K. W., Albieri, V., Dalton, S. O., Krøyer, A., Hove, H., Østergaard, J. R., Johansen, C., Sørensen, S. A., Mulvihill, J., Falck Winther, J. & Bidstrup, P. E., Dec 2024, In: Orphanet Journal of Rare Diseases. 19, 1, 441.
Research output: Contribution to journal › Journal article › Research › peer-review

The role of pathogenic TCF12 variants in children with coronal craniosynostosis-a systematic review with addition of two novel cases
Foss-Skiftesvik, J., Larsen, C. C., Stoltze, U. K., Kofod, T., Hove, H., Bøgeskov, L. & Østergaard, E., Nov 2024, In: Child's nervous system : ChNS : official journal of the International Society for Pediatric Neurosurgery. 40, 11, p. 3655-3671 17 p.
Research output: Contribution to journal › Review › peer-review

Medfødte misdannelser på hånd og fingre
Ottesen, C. S., Hove, H. & Weis, T., 5 Aug 2024, In: Ugeskrift for Laeger. 186, 32, V04230238.
Research output: Contribution to journal › Review › peer-review

Prevalence and Patient Characteristics of Ectodermal Dysplasias in Denmark
Herlin, L. K., Schmidt, S. A. J., Hermann, X. B., Rønholt, K., Bygum, A., Schuster, A., Lei, U., Mogensen, M., Vinding, G. R., Djursby, M., Hove, H., Blechingberg, J., Graversen, L., Mogensen, T. H., Gjørup, H., Langan, S. M. & Sommerlund, M., 1 May 2024, In: JAMA Dermatology. 160, 5, p. 502-510 9 p.
Research output: Contribution to journal › Journal article › Research › peer-review

Novel Alu insertion in the ZEB2 gene causing Mowat-Wilson syndrome
Barington, M., Bak, M., Kjartansdóttir, K. R., Hansen, T. V. O., Birkedal, U., Østergaard, E. & Hove, H. B., 2024, In: American Journal of Medical Genetics. Part A. 194, 8, e63581.
Research output: Contribution to journal › Journal article › Research › peer-review

Novel biallelic PISD missense variants cause spondyloepimetaphyseal dysplasia with disproportionate short stature and fragmented mitochondrial morphology
Aagaard Nolting, L., Holling, T., Nishimura, G., Ek, J., Bak, M., Ljungberg, M., Kutsche, K. & Hove, H., 2024, In: Clinical Genetics. 106, 3, p. 360-366 7 p.
Research output: Contribution to journal › Journal article › Research › peer-review

Employment, occupation, and income in adults with neurofibromatosis 1 in Denmark: a population- and register-based cohort study
Kenborg, L., Frederiksen, L. E., Galanakis, M., Doser, K., Nielsen, T. T., Doherty, M. A., Hove, H., Østergaard, J. R., Handrup, M. M., Ejerskov, C., Mulvihill, J. J. & Winther, J. F., 6 Nov 2023, In: Orphanet Journal of Rare Diseases. 18, 1, 346.
Research output: Contribution to journal › Journal article › Research › peer-review

Endocrine morbidity in neurofibromatosis 1: A nationwide, register-based cohort study
Kenborg, L., Ebbehoj, A., Ejerskov, C., Handrup, M. M., Østergaard, J. R., Hove, H., Doser, K., Krøyer, A., Mulvihill, J. J., Winther, J. F. & Stochholm, K., 2 Aug 2023, In: European Journal of Endocrinology. 189, 2, p. 190-198 9 p.

Research output: Contribution to journal › Journal article › Research › peer-review

School performance of children with neurofibromatosis 1: a nationwide population-based study
Doser, K., Belmonte, F., Andersen, K. K., Østergaard, J. R., Hove, H., Handrup, M. M., Ejerskov, C., Mulvihill, J. J., Winther, J. F. & Kenborg, L., Dec 2022, In: European journal of human genetics : EJHG. 30, 12, p. 1405-1412 8 p.
Research output: Contribution to journal › Journal article › Research › peer-review

Early diagnosis enabling precision medicine treatment in a young boy with PIK3R1-related overgrowth
Schönewolf-Greulich, B., Karstensen, H. G., Hjortshøj, T. D., Jørgensen, F. S., Harder, K. M., Frevert, S., Hove, H. & Diness, B. R., Oct 2022, In: European Journal of Medical Genetics. 65, 10, 104590.
Research output: Contribution to journal › Journal article › Research › peer-review

Cohort profile: life with neurofibromatosis 1 - the Danish NF1 cohort
Doser, K., Hove, H., Østergaard, J. R., Bidstrup, P. E., Dalton, S. O., Handrup, M. M., Ejerskov, C., Krøyer, A., Doherty, M. A., Møllegaard Jepsen, J. R., Mulvihill, J. J., Winther, J. F. & Kenborg, L., 20 Sept 2022, In: BMJ Paediatrics Open. 12, 9, p. e065340 e065340.
Research output: Contribution to journal › Journal article › Research › peer-review

Phenotypic spectrum of the recurrent TRPM3 p.(Val837Met) substitution in seven individuals with global developmental delay and hypotonia
Lines, M. A., Goldenberg, P., Wong, A., Srivastava, S., Bayat, A., Hove, H., Karstensen, H. G., Anyane-Yeboa, K., Liao, J., Jiang, N., May, A., Guzman, E., Morleo, M., D'Arrigo, S., Ciaccio, C., Pantaleoni, C., Castello, R., McKee, S., Ong, J. & Zibdeh-Lough, H. & 12 others, Tran-Mau-Them, F., Gerasimenko, A., Heron, D., Keren, B., Margot, H., de Sainte Agathe, J.-M., Burglen, L., Voets, T., Vriens, J., Innes, A. M., Dyment, D. A. & TUDP Study Group, Jun 2022, In: American Journal of Medical Genetics. Part A. 188, 6, p. 1667-1675 9 p.
Research output: Contribution to journal › Journal article › Research › peer-review

Pregnancy outcomes in women with neurofibromatosis 1: a Danish population-based cohort study
Kenborg, L., Boschini, C., Bidstrup, P. E., Dalton, S. O., Doser, K., Nielsen, T. T., Krøyer, A., Johansen, C., Frederiksen, K., Sørensen, S. A., Hove, H., Østergaard, J. R., Mulvihill, J. J. & Winther, J. F., Mar 2022, In: Journal of Medical Genetics. 59, 3, p. 237-242 6 p.
Research output: Contribution to journal › Journal article › Research › peer-review

Psychiatric disorders in individuals with neurofibromatosis 1 in Denmark: A nationwide register-based cohort study
Kenborg, L., Andersen, E. W., Duun-Henriksen, A. K., Jepsen, J. R. M., Doser, K., Dalton, S. O., Bidstrup, P. E., Krøyer, A., Frederiksen, L. E., Johansen, C., Østergaard, J. R., Hove, H., Sørensen, S. A., Riccardi, V. M., Mulvihill, J. J. & Winther, J. F., Dec 2021, In: American Journal of Medical Genetics. Part A. 185, 12, p. 3706-3716 11 p.
Research output: Contribution to journal › Journal article › Research › peer-review

Non-aortic cardiovascular disease in Marfan syndrome: a nationwide epidemiological study
Andersen, N. H., Groth, K. A., Berglund, A., Hove, H., Gravholt, C. H. & Stochholm, K., Jul 2021, In: Clinical research in cardiology : official journal of the German Cardiac Society. 110, 7, p. 1106-1115 10 p.
Research output: Contribution to journal › Journal article › Research › peer-review

Facial Asymmetry in Nonsyndromic and Muenke Syndrome-Associated Unicoronal Synostosis: A 3-Dimensional Study Based on Facial Surfaces Extracted From CT Scans
Öwall, L., Darvann, T. A., Hove, H. B., Heliövaara, A., Dunø, M., Kreiborg, S. & Hermann, N. V., Jun 2021, In: Cleft Palate-Craniofacial Journal. 58, 6, p. 687-696 10 p.
Research output: Contribution to journal › Journal article › Research › peer-review

Fracture Rates and Fracture Risk in Patients With Marfan Syndrome: A Nationwide Register-Based Cohort Study
Folkestad, L., Stochholm, K., Groth, K., Hove, H., Andersen, N. H. & Gravholt, C. H., May 2021, In: Journal of bone and mineral research : the official journal of the American Society for Bone and Mineral Research. 36, 5, p. 901-909 9 p.
Research output: Contribution to journal › Journal article › Research › peer-review

Threee dimensional facial asymmetry in non-syndromic and Muenke sydrome associated unicoronal synostosis
Öwall, L., Darvann, T. A., Hove, H. B., Heliövaara, A., Dunø, M., Kreiborg, S. & Hermann, N. V., 2021.

Research output: Contribution to conference › Conference abstract for conference › Research › peer-review

Bone geometry, density, and microarchitecture in the distal radius and tibia in adults with Marfan Syndrome assessed by HR-pQCT
Folkestad, L., Groth, K. A., Shanbhogue, V., Hove, H., Kyhl, K., Østergaard, J. R., Jørgensen, N. R., Andersen, N. H. & Gravholt, C. H., Dec 2020, In: Journal of bone and mineral research : the official journal of the American Society for Bone and Mineral Research. 35, 12, p. 2335-2344 10 p.
Research output: Contribution to journal › Journal article › Research › peer-review

Description of a family with X-linked oculo-auriculo-vertebral spectrum associated with polyalanine tract expansion in ZIC3
Trimouille, A., Tingaud-Sequeira, A., Lacombe, D., Duelund Hjortshøj, T., Kreiborg, S., Buciek Hove, H. & Rooryck, C., Oct 2020, In: Clinical Genetics. 98, 4, p. 384-389 6 p.
Research output: Contribution to journal › Journal article › Research › peer-review

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, p. 116897
Research output: Contribution to journal › Journal article › Research › peer-review

Forming and ending marital or cohabiting relationships in a Danish population-based cohort of individuals with neurofibromatosis 1
Kjaer, T. K., Andersen, E. W., Olsen, M., Kenborg, L., Bidstrup, P. E., Doser, K., Hove, H., Østergaard, J. R., Johansen, C. , Sørensen, S. A., Mulvihill, J. J., Winther, J. F. & Dalton, S. O., Aug 2020, In: European journal of human genetics : EJHG. 28, 8, p. 1028-1033 6 p.
Research output: Contribution to journal › Journal article › Research › peer-review

Unique skeletal manifestations in patients with Primrose syndrome
Arora, V., Leon, E., Diaz, J., Hove, H. B., Carvalho, D. R., Kurosawa, K., Nishimura, N., Nishimura, G., Saxena, R., Ferreira, C., Puri, R. D. & Verma, I. C., Aug 2020, In: European Journal of Medical Genetics. 63, 8, p. 103967
Research output: Contribution to journal › Journal article › Research › peer-review

Clinical characteristics and quality of life, depression, and anxiety in adults with neurofibromatosis type 1: A nationwide study
Doser, K., Andersen, E. W., Kenborg, L., Dalton, S. O., Jepsen, J. R. M., Krøyer, A., Østergaard, J., Hove, H., Sørensen, S. A., Johansen, C., Mulvihill, J., Winther, J. F. & Bidstrup, P. E., Jul 2020, In: American Journal of Medical Genetics. Part A. 182, 7, p. 1704-1715 12 p.
Research output: Contribution to journal › Journal article › Research › peer-review

Multisystem burden of neurofibromatosis 1 in Denmark: registry- and population-based rates of hospitalizations over the life span
Kenborg, L., Duun-Henriksen, A. K., Dalton, S. O., Bidstrup, P. E., Doser, K., Rugbjerg, K., Pedersen, C., Krøyer, A., Johansen, C., Andersen, K. K., Østergaard, J. R., Hove, H., Sørensen, S. A., Riccardi, V. M., Mulvihill, J. J. & Winther, J. F., Jun 2020, In: Genetics in medicine : official journal of the American College of Medical Genetics. 22, 6, p. 1069-1078 10 p.
Research output: Contribution to journal › Journal article › Research › peer-review

Novel Clinical and Radiological Findings in a Family with Autosomal Recessive Omodysplasia
Bayat, A., Dunø, M., Kirchhoff, M., Jørgensen, F. S., Nishimura, G. & Hove, H. B., Jun 2020, In: Molecular Syndromology. 11, 2, p. 83-89 7 p.
Research output: Contribution to journal › Journal article › Research › peer-review

Primrose syndrome: Characterization of the phenotype in 42 patients
Melis, D., Carvalho, D., Barbaro-Dieber, T., Espay, A. J., Gambello, M. J., Gener, B., Gerkes, E., Hitzert, M. M., Hove, H. B., Jansen, S., Jira, P. E., Lachlan, K., Menke, L. A., Narayanan, V., Ortiz, D., Overwater, E., Posmyk, R., Ramsey, K., Rossi, A. & Sandoval, R. L. & 10 others, Stumpel, C., Stuurman, K. E., Cordeddu, V., Turnpenny, P., Strisciuglio, P., Tartaglia, M., Unger, S., Waters, T., Turnbull, C. & Hennekam, R. C., Jun 2020, In: Clinical Genetics. 97, 6, p. 890-901 12 p.

Research output: Contribution to journal › Journal article › Research › peer-review

Defining the clinical phenotype of Saul-Wilson syndrome

Ferreira, C. R., Zein, W. M., Huryn, L. A., Merker, A., Berger, S. I., Wilson, W. G., Tiller, G. E., Wolfe, L. A., Merideth, M., Carvalho, D. R., Duker, A. L., Bratke, H., Haug, M. G., Rohena, L., Hove, H. B., Xia, Z.-J., Ng, B. G., Freeze, H. H., Gabriel, M. & Russi, A. H. S. & 11 others, Brick, L., Kozenko, M., Earl, D. L., Tham, E., Nishimura, G., Phillips, J. A., Gahl, W. A., Hamid, R., Jackson, A. P., Grigelioniene, G. & Bober, M. B., May 2020, In: Genetics in medicine : official journal of the American College of Medical Genetics. 22, 5, p. 857-866 10 p.

Research output: Contribution to journal › Journal article › Research › peer-review

Phenotypic presentations of Hajdu-Cheney syndrome according to age - 5 distinct clinical presentations

Graversen, L., Handrup, M. M., Irving, M., Hove, H., Diness, B. R., Risom, L., Svaneby, D., Aagaard, M. M., Vogel, I., Gjørup, H., Davidsen, M., Hellfritzsch, M. B., Lauridsen, E. & Gregersen, P. A., Feb 2020, In: European Journal of Medical Genetics. 63, 2, p. 103650

Research output: Contribution to journal › Journal article › Research › peer-review

The CHD4-related syndrome: a comprehensive investigation of the clinical spectrum, genotype-phenotype correlations, and molecular basis

Weiss, K., Lazar, H. P., Kurolap, A., Martinez, A. F., Paperna, T., Cohen, L., Smeland, M. F., Whalen, S., Heide, S., Keren, B., Terhal, P., Irving, M., Takaku, M., Roberts, J. D., Petrovich, R. M., Schrier Vergano, S. A., Kenney, A., Hove, H. , DeChene, E. & Quinonez, S. C. & 29 others, Colin, E., Ziegler, A., Rumple, M., Jain, M., Monteil, D., Roeder, E. R., Nugent, K., van Haeringen, A., Gambello, M., Santani, A., Medne, L., Krock, B., Skraban, C. M., Zackai, E. H., Dubbs, H. A., Smol, T., Ghoumid, J., Parker, M. J., Wright, M., Turnpenny, P., Clayton-Smith, J., Metcalfe, K., Kurumizaka, H., Gelb, B. D., Baris Feldman, H., Campeau, P. M., Muenke, M., Wade, P. A. & Lachlan, K., Feb 2020, In: Genetics in medicine : official journal of the American College of Medical Genetics. 22, 2, p. 389-397 9 p.

Research output: Contribution to journal › Journal article › Research › peer-review

Phenotypic variability in Muenke syndrome-observations from five Danish families

Öwall, L., Kreiborg, S., Dunø, M., Hermann, N. V., Darvann, T. A. & Hove, H., 2020, In: Coronary Artery Disease. 29, 1, p. 1-9 9 p.

Research output: Contribution to journal › Journal article › Research › peer-review

Validity of First-Time Diagnoses of Inherited Ichthyosis in the Danish National Patient Registry and the Danish Pathology Registry

Kristensen, M. H., Schmidt, S. A. J., Kibsgaard, L., Hove, H., Sommerlund, M. & Koppelhus, U., 2020, In: Clinical Epidemiology. 12, p. 651-657 7 p.

Research output: Contribution to journal › Journal article › Research › peer-review

Multiple Fractures and Impaired Bone Fracture Healing in a Patient with Pycnodysostosis and Hypophosphatasia

Hepp, N., Frederiksen, A. L., Dunø, M., Jørgensen, N. R., Langdahl, B., Hove, H. B., Vedtofte, P., Hindsø, K. & Jensen, J.-E. B., Dec 2019, In: Calcified Tissue International. 105, 6, p. 681-686 6 p.

Research output: Contribution to journal › Journal article › Research › peer-review

Correction: Educational delay and attainment in persons with neurofibromatosis 1 in Denmark

Doser, K., Kenborg, L., Andersen, E. W., Bidstrup, P. E., Kroyer, A., Hove, H., Østergaard, J., Sørensen, S. A., Johansen, C., Mulvihill, J., Winther, J. F. & Dalton, S. O., Jun 2019, 3 p.

Research output: Other contribution › Research

Novel de novo mutation in ZBTB20 in primrose syndrome in boy with short stature

Grímsdóttir, S., Hove, H. B., Kreiborg, S., Ek, J., Johansen, A., Darvann, T. A. & Hermann, N. V., Jan 2019, In: Clinical Dysmorphology. 28, 1, p. 41-45 5 p.

Research output: Contribution to journal › Journal article › Research › peer-review

Aortic aneurysm: An underestimated serious finding in the EP300 mutation phenotypical spectrum

Luyckx, I., Bolar, N., Diness, B. R., Hove, H. B., Verstraeten, A. & Loeys, B. L., 2019, In: European Journal of Medical Genetics. 62, 2, p. 96-96 1 p.

Research output: Contribution to journal › Journal article › Research › peer-review

Educational delay and attainment in persons with neurofibromatosis 1 in Denmark
Doser, K., Kenborg, L., Andersen, E. W., Bidstrup, P. E., Kroyer, A., Hove, H., Østergaard, J., Sørensen, S. A., Johansen, C., Mulvihill, J., Winther, J. F. & Dalton, S. O., 2019, In: European journal of human genetics : EJHG. 27, 6, p. 857-868 12 p.
Research output: Contribution to journal › Journal article › Research › peer-review

Spatially Detailed 3D Quantification of Improved Facial Symmetry After Surgery in Children With Unicoronal Synostosis
Öwall, L., Darvann, T. A., Hove, H. B., Bøgeskov, L., Kreiborg, S. & Hermann, N. V., 2019, In: Cleft Palate-Craniofacial Journal. 56, 7, p. 918-928 11 p.
Research output: Contribution to journal › Journal article › Research › peer-review

A Recurrent De Novo Heterozygous COG4 Substitution Leads to Saul-Wilson Syndrome, Disrupted Vesicular Trafficking, and Altered Proteoglycan Glycosylation
Ferreira, C. R., Xia, Z.-J., Clément, A., Parry, D. A., Davids, M., Taylan, F., Sharma, P., Turgeon, C. T., Blanco-Sánchez, B., Ng, B. G., Logan, C. V., Wolfe, L. A., Solomon, B. D., Cho, M. T., Douglas, G., Carvalho, D. R., Bratke, H., Haug, M. G., Phillips, J. B. & Wegner, J. & 31 others, Tiemeyer, M., Aoki, K., Nordgren, A., Hammarsjö, A., Duker, A. L., Rohena, L., Hove, H. B., Ek, J., Adams, D., Tifft, C. J., Onyekweli, T., Weixel, T., Macnamara, E., Radtke, K., Powis, Z., Earl, D., Gabriel, M., Russi, A. H. S., Brick, L., Kozenko, M., Tham, E., Raymond, K. M., Phillips, J. A., Tiller, G. E., Wilson, W. G., Hamid, R., Malicdan, M. C. V., Nishimura, G., Grigelioniene, G., Jackson, A. & Undiagnosed Diseases Network, 4 Oct 2018, In: American Journal of Human Genetics. 103, 4, p. 553-567 15 p.
Research output: Contribution to journal › Journal article › Research › peer-review

Causes of Mortality in the Marfan Syndrome(from a Nationwide Register Study)
Groth, K. A., Stochholm, K., Hove, H., Andersen, N. H. & Gravholt, C. H., 1 Oct 2018, In: The American journal of cardiology. 122, 7, p. 1231-1235 5 p.
Research output: Contribution to journal › Journal article › Research › peer-review

A study of familial Char syndrome involving the TFAP2B gene with a focus on facial shape characteristics
Nyboe, D., Kreiborg, S., Darvann, T., Dunø, M., Nissen, K. R. & Hove, H. B., Jul 2018, In: Clinical Dysmorphology. 27, 3, p. 71-77 7 p.
Research output: Contribution to journal › Journal article › Research › peer-review

Biomechanical properties of the patellar tendon in children with heritable connective tissue disorders
Jensen, J. K., Nygaard, R. H., Svensson, R. B., Hove, H. D., Magnusson, S. P., Kjær, M. & Couppé, C., Jul 2018, In: European Journal of Applied Physiology. 118, 7, p. 1301-1307 7 p.
Research output: Contribution to journal › Journal article › Research › peer-review

A complex phenotype in a family with a pathogenic SOX3 missense variant
Jelsig, A. M., Diness, B. R., Kreiborg, S., Main, K. M., Larsen, V. A. & Hove, H., 2018, In: European Journal of Medical Genetics. 61, 3, p. 168-72
Research output: Contribution to journal › Journal article › Research › peer-review

Improvement of Facial Symmetry after Surgery in Children with Unilateral Coronal Synostosis (UCS) Analyzed by Spatially Detailed 3D Quantification.
Öwall, B. L. C., Darvann, T. A., Hove, H. B., Hermann, N. V. & Kreiborg, S., 2018.
Research output: Contribution to conference › Conference abstract for conference › Research

Prenatal diagnosis of autosomal recessive Robinow syndrome using 3D ultrasound
Jeppesen, B. F., Hove, H. B., Kreiborg, S., Hermann, N. V., Darvann, T. A. & Jørgensen, F. S., Jul 2017, In: AACE clinical case reports. 5, 7, p. 1072-1076 5 p.
Research output: Contribution to journal › Journal article › Research › peer-review

En sjælden form for overvægt hos børn og unge
Christensen, S. Ø., Holm, K. & Hove, H. B., 20 Feb 2017, In: Ugeskrift for Laeger. 179, 11, p. 985
Research output: Contribution to journal › Journal article › Research › peer-review

Aortic events in a nationwide Marfan syndrome cohort
Groth, K. A., Stochholm, K., Hove, H., Kyhl, K., Gregersen, P. A., Vejlstrup, N., Østergaard, J. R., Gravholt, C. H. & Andersen, N. H., 2017, In: Clinical research in cardiology : official journal of the German Cardiac Society. 106, p. 105-112
Research output: Contribution to journal › Journal article › Research › peer-review

Spatially detailed 3D quantification of improvement of facial symmetry after surgery in children with Unilateral Coronal Synostosis (UCS).
Öwall, B. L. C., Darvann, T. A., Larsen, P., Hove, H. B., Hermann, N. V. & Kreiborg, S., 2017.
Research output: Contribution to conference › Conference abstract for conference › Research

Specific mosaic KRAS mutations affecting codon 146 cause oculoectodermal syndrome and encephalocraniocutaneous lipomatosis
Boppudi, S., Bögershausen, N., Hove, H. B., Percin, E. F., Aslan, D., Dvorsky, R., Kayhan, G., Li, Y., Cursiefen, C., Tantcheva-Poor, I., Toft, P. B., Bartsch, O., Lissewski, C., Wieland, I., Jakubiczka, S., Wollnik, B., Ahmadian, M. R., Heindl, L. M. & Zenker, M., Oct 2016, In: Clinical Genetics. 90, 4, p. 334-42 9 p.
Research output: Contribution to journal › Journal article › Research › peer-review

Acromelic frontonasal dysostosis and ZSWIM6 mutation: phenotypic spectrum and mosaicism
Twigg, S. R. F., Ousager, L. B., Miller, K. A., Zhou, Y., Elalaoui, S. C., Sefiani, A., Bak, G. S., Hove, H., Hansen, L. K., Fagerberg, C. R., Tajir, M. & Wilkie, A. O. M., Sept 2016, In: Clinical Genetics. 90, 3, p. 270-5 6 p.
Research output: Contribution to journal › Journal article › Research › peer-review

Monozygotic twins presenting with isolated sagittal and bicoronal synostosis, respectively
Hove, H. D., Dunø, M., Larsen, P. & Kreiborg, S., Apr 2016, In: Clinical Dysmorphology. 25, 2, p. 86-9 4 p.
Research output: Contribution to journal › Journal article › Research › peer-review

Facial Asymmetry in Children with Unicoronal Synostosis Who Have Undergone Craniofacial Reconstruction in Infancy
Öwall, B. L. C., Darvann, T. A., Larsen, P., Hove, H. D., Hermann, N. V., Bøgeskov, L. & Kreiborg, S., 2016, In: Cleft Palate - Craniofacial Journal. 53, 4, p. 385-93
Research output: Contribution to journal › Journal article › Research › peer-review

Monozygotic twins with a de novo 0.32Mb 16q24.3 deletion, including TUBB3 presenting with developmental delay and mild facial dysmorphism but without overt brain malformation
Grønborg, S., Kjaergaard, S., Hove, H., Larsen, V. A. & Kirchhoff, E. M., Nov 2015, In: American Journal of Medical Genetics. Part A. 167, 11, p. 2731-6 6 p.
Research output: Contribution to journal › Journal article › Research › peer-review

DVL1 frameshift mutations clustering in the penultimate exon cause autosomal-dominant Robinow syndrome
White, J., Mazzeu, J. F., Hoischen, A., Jhangiani, S. N., Gambin, T., Alcino, M. C., Penney, S., Saraiva, J. M., Hove, H., Skovby, F., Kayserili, H., Estrella, E., Vulto-van Silfhout, A. T., Steehouwer, M., Muzny, D. M., Sutton, V. R., Gibbs, R. A., Lupski, J. R., Brunner, H. G. & van Bon, B. W. M. & 2 others, Carvalho, C. M. B. & Baylor-Hopkins Center for Mendelian Genomics, 2 Apr 2015, In: American Journal of Human Genetics. 96, 4, p. 612-22 11 p.
Research output: Contribution to journal › Journal article › Research › peer-review

A study of the clinical and radiological features in a cohort of 93 patients with a COL2A1 mutation causing spondyloepiphyseal dysplasia congenita or a related phenotype
Terhal, P. A., Nievelstein, R. J. A. J., Verver, E. J. J., Topsakal, V., van Dommelen, P., Hoornaert, K., Le Merrer, M., Zankl, A., Simon, M. E. H., Smithson, S. F., Marcelis, C., Kerr, B., Clayton-Smith, J., Kinning, E., Mansour, S., Elmslie, F., Goodwin, L., van der Hout, A. H., Veenstra-Knol, H. E. & Herkert, J. C. & 36 others, Lund, A. M., Hennekam, R. C. M., Mégarbané, A., Lees, M. M., Wilson, L. C., Male, A., Hurst, J., Alanay, Y., Annerén, G., Betz, R. C., Bongers, E. M. H. F., Cormier-Daire, V., Dieux, A., David, A., Elting, M. W., van den Ende, J., Green, A., van Hagen, J. M., Hertel, N. T., Holder-Espinasse, M., den Hollander, N., Homfray, T., Hove, H. D., Price, S., Raas-Rothschild, A., Rohrbach, M., Schroeter, B., Suri, M., Thompson, E. M., Tobias, E. S., Toutain, A., Vreeburg, M., Wakeling, E., Knoers, N. V., Coucke, P. & Mortier, G. R., Mar 2015, In: American Journal of Medical Genetics. Part A. 167, 3, p. 461-75 15 p.
Research output: Contribution to journal › Journal article › Research › peer-review

Familial craniosynostosis associated with a microdeletion involving the NFIA gene
Nyboe, D., Kreiborg, S., Kirchhoff, E. M. & Hove, H. B., 24 Feb 2015, In: Clinical Dysmorphology. 24, 3, p. 109-12
Research output: Contribution to journal › Journal article › Research › peer-review

Prevalence, incidence, and age at diagnosis in Marfan Syndrome
Groth, K., Hove, H., Kyhl, K., Folkestad, L., Gaustadnes, M., Vejlstrup, N., Stochholm, K., Østergaard, J. R., Andersen, N. & Gravholt, C. H., 2015, In: Orphanet Journal of Rare Diseases. 10, p. 153
Research output: Contribution to journal › Journal article › Research › peer-review

Using CT to validate measurement of facial asymmetry in surface scans of infants with unilateral coronal synostosis.
Darvann, T. A., Öwall, B. L. C., Hermann, N. V., Larsen, P., Hove, H. B. & Kreiborg, S., 2015.
Research output: Contribution to conference › Conference abstract for conference › Research

The prevalence of CHD7 missense versus truncating mutations is higher in patients with Kallmann syndrome than in typical CHARGE patients
Marcos, S., Sarfati, J., Leroy, C., Fouveaut, C., Parent, P., Metz, C., Wolczynski, S., Gérard, M., Bieth, E., Kurtz, F., Verier-Mine, O., Perrin, L., Archambeaud, F., Cabrol, S., Rodien, P., Hove, H., Prescott, T., Lacombe, D., Christin-Maitre, S. & Touraine, P. & 6 others, Hieronimus, S., Dewailly, D., Young, J., Pugeat, M., Hardelin, J.-P. & Dodé, C., Oct 2014, In: The Journal of clinical endocrinology and metabolism. 99, 10, p. E2138-43
Research output: Contribution to journal › Journal article › Research › peer-review

Facial Asymmetry in Children with Unicoronal Synostosis Who Have Undergone Craniofacial Reconstruction in Infancy.
Öwall, B. L. C., Darvann, T. A., Larsen, P., Hove, H. B., Hermann, N. V., Bøgeskov, L. & Kreiborg, S., 2014.
Research output: Contribution to conference › Conference abstract for conference › Research

Delineation of a new chromosome 20q11.2 duplication syndrome including the ASXL1 gene
Avila, M., Kirchhoff, E. M., Marle, N., Hove, H. D., Chouchane, M., Thauvin-Robinet, C., Masurel, A., Mosca-Boidron, A.-L., Callier, P., Mugneret, F., Kjaergaard, S. & Faivre, L., Jul 2013, In: American Journal of Medical Genetics. Part A. 161A , 7, p. 1594-8 5 p.
Research output: Contribution to journal › Journal article › Research › peer-review

The cardiac phenotype in patients with a CHD7 mutation
Corsten-Janssen, N., Kerstjens-Frederikse, W. S., du Marchie Sarvaas, G. J., Baardman, M. E., Bakker, M. K., Bergman, J. E. H., Hove, H. D., Heimdal, K. R., Rustad, C. F., Hennekam, R. C. M., Hofstra, R. M. W., Hoefsloot, L. H., Van Ravenswaaij-Arts, C. M. A. & Kapusta, L., Jun 2013, In: Circulation. Cardiovascular genetics. 6, 3, p. 248-54 7 p.
Research output: Contribution to journal › Journal article › Research › peer-review

Børn med hypermobilitet
Juul-Kristensen, B., Hove, H. D. & Remvig, L., 18 Mar 2013, *Pædiatrisk Fysioterapi*. Harboe, H. & Stegger, H. (eds.). 1. ed. Akademisk Forlag, p. 205-222
Research output: Chapter in Book/Report/Conference proceeding › Book chapter › Education

Novel mutations including deletions of the entire OFD1 gene in 30 families with type 1 orofaciodigital syndrome: a study of the extensive clinical variability
Bisschoff, I. J., Zeschnigk, C., Horn, D., Wellek, B., Rieß, A., Wessels, M., Willems, P., Jensen, P., Busche, A., Bekkebraten, J., Chopra, M., Hove, H. D., Evers, A. C. W., Heimdal, K., Kaiser, A.-S., Kunstmann, E., Robinson, K. L., Linné, M., Martin, P. & McGrath, J. & 10 others, Pradel, W., Prescott, K. E., Roesler, B., Rudolf, G., Siebers-Renelt, U., Tyshchenko, N., Wieczorek, D., Wolff, G., Dobyns, W. B. & Morris-Rosendahl, D. J., Jan 2013, In: Human Mutation. 34, 1, p. 237-47 11 p.
Research output: Contribution to journal › Journal article › Research › peer-review

Identification of six novel PTH1R mutations in families with a history of primary failure of tooth eruption
Risom, L., Christoffersen, L., Daugaard-Jensen, J., Hove, H. D., Andersen, H. S., Andresen, B. S., Kreiborg, S. & Duno, M., 2013, In: P L o S One. 8, 9, p. e74601
Research output: Contribution to journal › Journal article › Research › peer-review

Osteopathia striata congenita with cranial sclerosis and intellectual disability due to contiguous gene deletions involving the WTX locus
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Research output: Contribution to journal › Journal article › Research › peer-review

Phenotype in 18 Danish subjects with genetically verified CHARGE syndrome
Husu, E., Hove, H. B., Farholt, S., Bille, M., Tranebjaerg, L., Vogel, I. & Kreiborg, S., 2013, In: Clinical Genetics. 83, 2, p. 125-134 10 p.
Research output: Contribution to journal › Journal article › Research › peer-review

Crouzon syndrome associated with acanthosis nigricans: prenatal 2D and 3D ultrasound findings and postnatal 3D CT findings
Nørgaard, P., Hagen, C. P., Hove, H., Dunø, M., Nissen, K. R., Kreiborg, S. & Jørgensen, F. S., 2012, In: Acta Radiologica Short Reports. 1, 4
Research output: Contribution to journal › Journal article › Research › peer-review

Crouzon syndrome associated with acanthosis nigricans: prenatal 2D and 3D ultrasound findings and postnatal 3D CT findings
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Microdeletion in distal 17p13.1: a recognizable phenotype with microcephaly, distinctive facial features, and intellectual disability
Zeesman, S., Kjaergaard, S., Hove, H. B., Kirchhoff, E. M., Stevens, J. M. & Nowaczyk, M. J. M., 2012, In: American Journal of Medical Genetics. Part A. 158A, 8, p. 1832-6 5 p.
Research output: Contribution to journal › Journal article › Research › peer-review

Plexiform neurofibroma of the eye region occurring in patients without neurofibromatosis type 1
Bechtold, D., Hove, H. B., Prause, J. U., Heegaard, S. & Toft, P. B., 2012, In: Ophthalmic Plastic and Reconstructive Surgery. 28, 6, p. 413-5 3 p.
Research output: Contribution to journal › Journal article › Research › peer-review

Bohring-Opitz (Oberklaid-Danks) syndrome: clinical study, review of the literature, and discussion of possible pathogenesis
Hastings, R., Cobben, J.-M., Gillessen-Kaesbach, G., Goodship, J., Hove, H. B., Kjærgaard, S., Kemp, H., Kingston, H., Lunt, P., Mansour, S., McGowan, R., Metcalfe, K., Murdoch-Davis, C., Ray, M., Rio, M., Smithson, S., Tolmie, J., Turnpenny, P., van Bon, B. & Wieczorek, D. & 1 others, Newbury-Ecob, R., 2011, In: European Journal of Human Genetics. 19, 5, p. 513-9 7 p.
Research output: Contribution to journal › Journal article › Research › peer-review

Deficiency of the cytoskeletal protein SPECC1L leads to oblique facial clefting
Saadi, I., Alkuraya, F. S., Gisselbrecht, S. S., Goessling, W., Cavallesco, R., Turbe-Doan, A., Petrin, A. L., Harris, J., Siddiqui, U., Grix, A. W., Hove, H. B., Leboulch, P., Glover, T. W., Morton, C. C., Richieri-Costa, A., Murray, J. C., Erickson, R. P. & Maas, R. L., 2011, In: American Journal of Human Genetics. 89, 1, p. 44-55 12 p.
Research output: Contribution to journal › Journal article › Research › peer-review

RUNX2 analysis of Danish cleidocranial dysplasia families
Hansen, L., Riis, A. K., Silahtaroglu, A., Hove, H. B., Lauridsen, E., Eiberg, H. R. L. & Kreiborg, S., 2011, In: Clinical Genetics. 79, 3, p. 254-63 10 p.
Research output: Contribution to journal › Journal article › Research › peer-review

Skin signs in Ehlers-Danlos syndrome: clinical tests and para-clinical methods
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Research output: Contribution to journal › Journal article › Research › peer-review

A 15q24 microduplication, reciprocal to the recently described 15q24 microdeletion, in a boy sharing clinical features with 15q24 microdeletion syndrome patients
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Research output: Contribution to journal › Journal article › Research › peer-review

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Research output: Contribution to journal › Journal article › Research › peer-review

An echo-poor spine at 13 weeks: an early sign of cleidocranial dysplasia
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Craniofacial morphology in Muenke syndrome
Keller, M. K., Hermann, N. V., Darvann, T. A., Larsen, P., Hove, H. D., Christensen, L., Schwartz, M., Marsh, J. L. & Kreiborg, S., Mar 2007, In: The Journal of craniofacial surgery. 18, 2, p. 374-86 13 p.
Research output: Contribution to journal › Journal article › Research › peer-review

Analysis of 65 tuberous sclerosis complex (TSC) patients by TSC2 DGGE, TSC1/TSC2 MLPA, and TSC1 long-range PCR sequencing, and report of 28 novel mutations
Rendtorff, N. D., Bjerregaard, B., Frödin, M., Kjaergaard, S., Hove, H., Skovby, F., Brøndum-Nielsen, K., Schwartz, M. & Danish Tuberous Sclerosis Group, Oct 2005, In: Human Mutation. 26, 4, p. 374-83 10 p.
Research output: Contribution to journal › Journal article › Research › peer-review

Mapping genomic deletions down to the base: a quantitative copy number scanning approach used to characterise and clone the breakpoints of a recurrent 7p14.2p15.3 deletion
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Research output: Contribution to journal › Journal article › Research › peer-review

Svaer obstruktiv søvnapnø hos et barn med kraniofacial anomali
Qvist, J., Hove, H. D., Welling, K.-L. K. & Kreiborg, S., 16 Aug 2004, In: Ugeskrift for Laeger. 166, 34, p. 2910-2 3 p.
Research output: Contribution to journal › Journal article › Research › peer-review

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Research output: Contribution to journal › Journal article › Research › peer-review

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Research output: Contribution to journal › Review › peer-review

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Hove, H., Feb 1998, In: Danish Medical Journal. 45, 1, p. 15-33 19 p.
Research output: Contribution to journal › Review › peer-review

Colonic production of butyrate in patients with previous colonic cancer during long-term treatment with dietary fibre (Plantago ovata seeds)
Nordgaard, I., Hove, H., Clausen, M. R. & Mortensen, P. B., Oct 1996, In: Scandinavian Journal of Gastroenterology. 31, 10, p. 1011-20 10 p.
Research output: Contribution to journal › Journal article › Research › peer-review

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Research output: Contribution to journal › Journal article › Research › peer-review

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Research output: Contribution to journal › Journal article › Research › peer-review

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Research output: Contribution to journal › Journal article › Research › peer-review

Faecal DL-lactate concentration in 100 gastrointestinal patients
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Research output: Contribution to journal › Journal article › Research › peer-review

Effect of lactic acid bacteria on the intestinal production of lactate and short-chain fatty acids, and the absorption of lactose
Hove, H., Nordgaard-Andersen, I. & Mortensen, P. B., Jan 1994, In: The American journal of clinical nutrition. 59, 1, p. 74-9 6 p.
Research output: Contribution to journal › Journal article › Research › peer-review

Anti-cardiolipin antibodies (IgG and IgA) in women with recurrent fetal loss correlate to clinical and serological characteristics of SLE
Bagger, P. V., Andersen, V., Baslund, B., Beck, B., Hove, H., Høier-Madsen, M., Petersen, J., Philip, J., Schaadt, O. & Skouby, S. O., Aug 1993, In: Acta Obstetricia et Gynecologica Scandinavica. 72, 6, p. 465-9 5 p.
Research output: Contribution to journal › Journal article › Research › peer-review

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Research output: Contribution to journal › Journal article › Research › peer-review

Colonic fermentation of ispaghula, wheat bran, glucose, and albumin to short-chain fatty acids and ammonia evaluated in vitro in 50 subjects
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Research output: Contribution to journal › Journal article › Research › peer-review

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Holtug, K., Clausen, M. R., Hove, H., Christiansen, J. & Mortensen, P. B., Jul 1992, In: Scandinavian Journal of Gastroenterology. 27, 7, p. 545-52 8 p.
Research output: Contribution to journal › Journal article › Research › peer-review

Fermentation to short-chain fatty acids and lactate in human faecal batch cultures. Intra- and inter-individual variations versus variations caused by changes in fermented saccharides
Mortensen, P. B., Hove, H., Clausen, M. R. & Holtug, K., Dec 1991, In: Scandinavian Journal of Gastroenterology. 26, 12, p. 1285-94 10 p.
Research output: Contribution to journal › Journal article › Research › peer-review

Activities

Achondroplasia - an up-date
Hove, H. B. (Lecturer)
21 Jan 2019

Prader-Willi syndrom
Hove, H. B. (Lecturer)
6 Apr 2018

Nordic Skeletal Dysplasia Symposium
Hove, H. B. (Organizer)
8 Mar 2018 → 9 Mar 2018

Nordic network for Rare Disease (External organisation)
Hove, H. B. (Member)
2017 → 2019

4th Nordic Conference on Rare Diseases.
Hove, H. B. (Lecturer)
Sept 2016

National Strategie for Sjældne Sygdomme
Hove, H. B. (Consultant)
2012 → 2014