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Research interests

Rare Diseases, Dysmorphology, Genomic Medicine, Genetic Diagnostics of Rare Diseases and Syndromes with or without Intellectual Disability, Clinical Genetics, and Genetic Counselling.

Employment

Department of Clinical Research

1. Jun 2021 → 31. Jul 2025

Clinical Professor

KI, OUH, Research unit of Clinical Genetics (Odense)

1. Jun 2021 → 31. Jul 2025

Head of Department

Odense University Hospital

Odense, Denmark

1. Jul 2022 → present

Head of Department

Odense University Hospital

Odense, Denmark

1. Mar 2010 → 30. Jun 2022

Research outputs

Late diagnosis of partial 3β-hydroxysteroid dehydrogenase type 2 deficiency - characterization of a new genetic variant

Øzdemir, C. M., Nielsen, M. M., Liimatta, J., Voegel, C. D., Elzenaty, R. N., Wasehuus, V. S., Lind-Holst, M., Ornstrup, M. J., Gram, S. B., Ousager, L. B., Flück, C. E. & Gravholt, C. H., 1. Jul 2024, In: Endocrinology, Diabetes & Metabolism Case Reports. 2024, 3

Comprehensive Noninvasive Fetal Screening by Deep Trio-Exome Sequencing

Miceikaitė, I., Hao, Q., Brasch-Andersen, C., Fagerberg, C. R., Torring, P. M., Kristiansen, B. S., Ousager, L. B., Sperling, L., Ibsen, M. H., Löser, K. & Larsen, M. J., 1. May 2024, In: Obstetrical and Gynecological Survey. 79, 5, p. 261-263

Identification of a founder variant AAGAB c.370C>T, p.Arg124Ter in patients with punctate palmoplantar keratoderma in Southern Denmark

Gram, S. B., Jørgensen, A. S. F., Bygum, A., Brusgaard, K. & Ousager, L. B., May 2024, In: Clinical Genetics. 105, 5, p. 561-566

Plantar keratoderma and curly hair as a diagnostic clue of cardiomyopathy risk

Gram, S. B., Brusgaard, K., Bygum, A., Christensen, A. H. & Ousager, L. B., May 2024, In: Journal of Dermatology. 51, 5, p. e143-e144

Risk of Cancer and Mortality in Peutz-Jeghers Syndrome and Juvenile Polyposis Syndrome—A Nationwide Cohort Study With Matched Controls

Jelsig, A. M., Wullum, L., Kuhlmann, T. P., Ousager, L. B., Burisch, J. & Karstensen, J. G., Dec 2023, In: Gastroenterology. 165, 6, p. 1565-1567.e2

Comprehensive Noninvasive Fetal Screening by Deep Trio-Exome Sequencing
Miceikaitė, I., Hao, Q., Brasch-Andersen, C., Fagerberg, C. R., Torring, P. M., Kristiansen, B. S., Ousager, L. B., Sperling, L., Ibsen, M. H., Löser, K. & Larsen, M. J., 23. Nov 2023, In: The New England Journal of Medicine. 389, 21, p. 2017-2019

Structural and non-coding variants increase the diagnostic yield of clinical whole genome sequencing for rare diseases
Pagnamenta, A. T., Camps, C., Giacopuzzi, E., Taylor, J. M., Hashim, M., Calpena, E., Kaisaki, P. J., Hashimoto, A., Yu, J., Sanders, E., Schwessinger, R., Hughes, J. R., Lunter, G., Dreau, H., Ferla, M., Lange, L., Kesim, Y., Ragoussis, V., Vavoulis, D. V. & Allroggen, H. & 77 others, Ansorge, O., Babbs, C., Banka, S., Baños-Piñero, B., Beeson, D., Ben-Ami, T., Bennett, D. L., Bento, C., Blair, E., Brasch-Andersen, C., Bull, K. R., Cario, H., Cilliers, D., Conti, V., Davies, E. G., Dhalla, F., Dacal, B. D., Dong, Y., Dunford, J. E., Guerrini, R., Harris, A. L., Hartley, J., Hollander, G., Javaid, K., Kane, M., Kelly, D., Kelly, D., Knight, S. J. L., Kreins, A. Y., Kvikstad, E. M., Langman, C. B., Lester, T., Lines, K. E., Lord, S. R., Lu, X., Mansour, S., Manzur, A., Maroofian, R., Marsden, B., Mason, J., McGowan, S. J., Mei, D., Mlcochova, H., Murakami, Y., Németh, A. H., Okoli, S., Ormondroyd, E., Ousager, L. B., Palace, J., Patel, S. Y., Pentony, M. M., Pugh, C., Rad, A., Ramesh, A., Riva, S. G., Roberts, I., Roy, N., Salminen, O., Schilling, K. D., Scott, C., Sen, A., Smith, C., Stevenson, M., Thakker, R. V., Twigg, S. R. F., Uhlig, H. H., van Wijk, R., Vona, B., Wall, S., Wang, J., Watkins, H., Zak, J., Schuh, A. H., Kini, U., Wilkie, A. O. M., Popitsch, N. & Taylor, J. C., 9. Nov 2023, In: Genome Medicine. 15, 25 p., 94.

Cancer risk and mortality in patients with solitary juvenile polyps-A nationwide cohort study with matched controls
Jelsig, A. M., Wullum, L., Kuhlmann, T. P., Ousager, L. B., Burisch, J. & Karstensen, J. G., Oct 2023, In: United European Gastroenterology Journal. 11, 8, p. 745-749

Cancer risk and mortality in patients with solitary Peutz-Jeghers polyps
Jelsig, A. M., Wullum, L., Ousager, L. B., Burish, J., Kühlmann, T. P. & Karstensen, J. G., 22. Sept 2023, In: Gastroenterology Report. 11, goad056.

Is punctate palmoplantar keratoderma type 1 associated with malignancy? A systematic review of the literature
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Comprehensive prenatal diagnostics: Exome versus genome sequencing
Miceikaite, I., Fagerberg, C., Brasch-Andersen, C., Torring, P. M., Kristiansen, B. S., Hao, Q., Sperling, L., Ibsen, M. H., Löser, K., Bendsen, E. A., Ousager, L. B. & Larsen, M. J., Aug 2023, In: Prenatal Diagnosis. 43, 9, p. 1132-1141

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Jelsig, A. M., van Overeem Hansen, T., Gede, L. B., Qvist, N., Christensen, L.-L., Lautrup, C. K., Frederiksen, J. H., Sunde, L., Ousager, L. B., Ljungmann, K., Bertelsen, B. & Karstensen, J. G., Jul 2023, In: Clinical Genetics. 104, 1, p. 81-89

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Gram, S. B., Brusgaard, K., Ousager, L. B. & Bygum, A., 27. Feb 2023, (E-pub ahead of print) In: Ugeskrift for Læger. V71166.

Further clinical and molecular characterization of an XLID syndrome associated with BRWD3 variants, a gene implicated in the leukemia-related JAK-STAT pathway
Delanne, J., Lecat, M., Blackburn, P. R., Klee, E. W., Stumpel, C. T. R. M., Stegmann, S., Stevens, S. J. C., Nava, C., Heron, D., Keren, B., Mahida, S., Naidu, S., Babovic-Vuksanovic, D., Herkert, J. C., Torring, P. M., Kibæk, M., De Bie, I., Pfundt, R., Hendriks, Y. M. C. & Ousager, L. B. & 15 others, Bend, R., Warren, H., Skinner, S. A., Lyons, M. J., Pöe, C., Chevarin, M., Jouan, T., Garde, A., Thomas, Q., Kuentz, P., Tisserant, E., Duffourd, Y., Philippe, C., Faivre, L. & Thauvin-Robinet, C., Jan 2023, In: European Journal of Medical Genetics. 66, 1, 7 p., 104670.

A novel heterozygote LORICRIN variant in a father and daughter with palmoplantar keratoderma
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Identification of a Novel PLCD1 Variant in a Danish Family with Hereditary Leukonychia
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Familial Multiple Lipomatosis - analyses of genetic etiology by whole genome sequencing and delineation of the clinical phenotype
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Klinisk genetik: Genetisk udredning og rådgivning
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Klinisk genetik: Etik og jura
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Sygdommes genetik: Syndromer, medfødte misdannelser og mental retardering
Kibæk Nielsen, I. & Ousager, L. B., 2022, *Medicinsk genetik.* Østergaard, E. & Sunde, L. (eds.). 3. ed. FADL's Forlag, p. 393-422 16

Truncating SRCAP variants outside the Floating-Harbor syndrome locus cause a distinct neurodevelopmental disorder with a specific DNA methylation signature
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Chromosomal translocation disrupting the SMAD4 gene resulting in the combined phenotype of Juvenile polyposis syndrome and Hereditary Hemorrhagic Telangiectasia
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Brusgaard, K., Torring, P. M., Kjeldsen, A. D. & Ousager, L. B., 1. Feb 2018, In: Angiogenesis. 21, 1, p. 146-147

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Press/Media

Erfaren overlæge bliver professor i genetik
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02/09/2020
1 Media contribution

Fagfolk frygter slingrekurs på vejen mod personlig medicin
Ousager, L. B.
04/08/2023
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TV-2 Fyn article 24th of Jan. 2020 'Nyt center skal skabe tryghed hos patienter med sjældne sygdomme'
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25/01/2020
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TV-2 Fyn Nyheder 25th of Jan. 2020
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