

List of publications for Benedicte Paus

Name prior to 16.11.01: Benedicte Christensen

Updated 25.02.2025

Original publications in scientific journals with peer review

1 Ueland PM, Refsum HM, **Christensen B**

Methotrexate sensitivity in Down syndrome: A hypothesis
Cancer Chemotherapy and Pharmacology (1990) 25: 384-6

2 **Christensen B**, Refsum H, Vintermyr O, Ueland PM

Homocysteine export from cells cultured in the presence of physiological or superfuous levels of methionine: methionine loading of non-transformed, transformed, proliferating and quiescent cells in culture
Journal of Cell Physiology (1991) 146: 52-62

3 **Christensen B**, Lillehaug JR, Ueland PM

Interaction between methotrexate, „rescue“ agents and cell proliferation as modulators of homocysteine export from cells in culture
The Journal of Pharmacology and Experimental Therapeutics (1991) 258: 559-566

4 Garras A, Djurhuus R, **Christensen B**, Lillehaug JR, Ueland PM

A non-radioactive assay for N5-methyltetrahydrofolate-reductase-homocysteine methyltransferase (methionine synthase) based on o-phthaldialdehyde derivatization of methionine and fluorescence detection
Analytical Biochemistry (1991) 199: 112-118

5 **Christensen B**, Refsum H, Garras A, Ueland PM

Homocysteine remethylation during nitrous oxide exposure of cells cultured in media containing various concentrations of folates
The journal of Pharmacology and Experimantal Therapeutics (1992) 261(3): 1096-1105

6 Warren DJ, **Christensen B**, Slørdal L

Effect of nitrous oxide on hematopoiesis in vitro: Biochemical and functional features
Pharmacology and Toxicology (1993) 72: 69-72

7 **Christensen B** and Ueland PM

Methionine synthase inactivation by nitrous oxide during methionine loading of normal human fibroblasts. Homocysteine remethylation as determinant of enzyme inactivation and homocysteine export
The Journal of Pharmacology and Experimental Therapeutics (1993) 267(3):1298-1303

8 **Christensen B**, Rosenblatt DS, Chu R, and Ueland PM

Effect of methionine and nitrous oxide on homocysteine export and remethylation in fibroblasts from cystathionine synthase deficient, cblG and cblE patients

Pediatric Research (1994) 35(1): 3-9

9 **Christensen B**, Guttormsen AB, Schneede J, Refsum H, Riedel B, Svardal A, Ueland PM
Preoperative methionine loading enhances restoration of the cobalamin-dependent enzyme
methionine synthase after nitrous oxide anaesthesia

Anaesthesiology (1994) 80(5): 1046-1056

10 Fiskerstrand T, **Christensen B**, Tysnes OB, Ueland PM, Refsum H
Development and reversion of methionine dependence in a human glioma cell line: Relation to
homocysteine remethylation and cobalamin status

Cancer Research (1994) 54: 4899-4906

11 Riudor E, Ribes A, Pérez-Cerdá C, Arranz JA, Mora J, Yeste D, Castello F, **Christensen B**,
Søvik O

Metabolic coma with ketoacidosis and hyperglycemia in 2-methylacetoacetyl-CoA thiolase
deficiency

The Journal of Inherited Metabolic Disease (1995) 18(6): 748-749

12 Goyette P, **Christensen B**, Rosenblatt DS, Rozen R

Severe and mild mutations in cis for the methylenetetrahydrofolate reductase (MTHFR) gene,
and description of 5 novel mutations in MTHFR

American Journal of Human Genetics (1996) 59: 1268-1275

13 Leclerc D, Campeau E, Goyette P, Adjalla CE, **Christensen B**, Ross M, Eydoux P,
Rosenblatt DS, Rozen R, Gravel RA

Human methionine synthase: cDNA cloning, chromosomal localization, and identification of
mutations in patients of the cblG complementation group of folate/cobalamin disorders

Human Molecular Genetics (1996) 5(12): 1867-1874

14 **Christensen B**, Frosst P, Lussier-Cacan S, Selhub J, Rosenblatt DS, Genest J Jr, Rozen R
Correlation of a common mutation in methylenetetrahydrofolate reductase (MTHFR) with
plasma homocysteine in patients with premature coronary artery disease

Arteriosclerosis, Thrombosis and Vascular Biology (1997) 17(3): 569-573

15 Rand-Hendriksen S and **Christensen B**

Magnettomografi av sentralnervesystemet hos voksne med myelomeningocele

The Journal of the Norwegian Medical Association (1998) 118(27): 4208-10

16 Weisberg I, Tran P, **Christensen B**, Rozen R

A second genetic polymorphism in methylenetetrahydrofolate reductase (MTHFR) associated
with decreased enzyme activity

Molecular Genetics and Metabolism (1998) 64: 169-172

17 **Christensen B**, Arbour L, Tran P, Leclerc D, Sabaghian N, Platt R, Gilfix BM,
Rozenblatt DS, Gravel R, Forbes P, Rozen R

Genetic polymorphisms in methylenetetrahydrofolate reductase and methionine synthase, folate levels in red blood cells, and risk of neural tube defects
American Journal of Medical Genetics (1999) 84: 151-157

18 Ma J, Stampfer MJ, **Christensen B**, Giovannucci E, Hunter DJ, Chen J, Willett WC, Selhub J, Hennekens CH, Gravel R, Rozen R
A polymorphism of the methionine synthase gene: association with plasma folate, B12, homocyst(e)ine, and colorectal cancer risk
Cancer Epidemiology, Biomarkers & Prevention (1999) 8(9): 825-829

19 **Christensen B**, Landaas S, Stensvold I, Djurovic S, Retterstøl L, Ringstad J, Berg K, Thelle DS
Whole blood folate, homocysteine in serum, and risk of first acute myocardial infarction
Atherosclerosis (1999) 147: 317-326

20 Wilson A, Platt R, Wu Q, Leclerc D, **Christensen B**, Yang H, Gravel RA, Rozen R
A common variant in methionine synthase reductase combined with low cobalamin (vitamin B12) increases risk for spina bifida
Molecular Genetics and Metabolism (1999) 67: 317-323

21 Djurovic S, Thelle DS, Ringstad J, **Christensen B**, Berg K
Altered serum concentrations of TGF- β 1 and Lp(a) lipoproteins and their correlation in patients with first acute myocardial infarction
Nutrition, Metabolism and Cardiovascular Diseases (1999) 9: 250-254

22 Sibani S, **Christensen B**, O'Ferrel E, Saadi I, Hiou-Tim F, Rosenblatt DS, Rozen R
Characterization of six novel mutations in the methylenetetrahydrofolate reductase (MTHFR) gene in patients with homocystinuria
Human Mutation (2000) 15(3): 280-287

23 **Christensen B**, Blaas H-G, Isaksen CV, Roald B, Ørstavik KH
Sibs with anencephaly, anophthalmia, clefts, omphalocele, and polydactyly: Hydrolethalus or acrocallosal syndrome?
American Journal of Medical Genetics (2000) 91: 231-234

24 Omland T, Samuelsson A, Hartford M, Herlitz J, Karlsson T, **Christensen B**, Caidahl K
Serum homocysteine level as an indicator of survival in patients with acute coronary syndromes
Archives of Internal Medicine (2000) 160(12): 1834-40

25 **Christensen B**
Which antibiotics are appropriate to treat bacteriuria of pregnancy?
Journal of Antimicrobial Chemotherapy (2000) 46(Suppl.S1): 29-34

26 Holven K, Holm T, Aukrust P, Brosstad F, **Christensen B**, Kjekshus J, Andreassen AK, Gullestad L, Ose L, Svilaas A, Nenseter MS

Improved peripheral endothelial function by folic acid treatment in patients with hyperhomocysteinemia

The American Journal of Medicine (2001) 110: 536-542

27 Christensen B

Use of antibiotics to treat bacteriuria of pregnancy in the Nordic countries. Which antibiotics are appropriate to treat bacteriuria of pregnancy?

International Journal of Antimicrobial Agents (2001) 17: 283-285

28 Christensen B, Mosdol A, Retterstol L, Landaas S, Thelle DS. Abstention from filtered coffee reduces the levels of homocysteine and cholesterol – a randomized, controlled trial.

The American Journal of Clinical Nutrition (2001) 74(3): 302-307

29 Schumacher A, Seljeflot I, Sommervoll L, **Christensen B**, Otterstad JE, Arnesen H
Increased levels of endothelial haemostatic markers in patients with coronary heart disease

Thrombosis Research (2002) 105(4): 25-31

30 Schumacher A, Seljeflot I, Sommervoll L, **Christensen B**, Otterstad JE, Arnesen H
Increased levels of markers of vascular inflammation in patients with coronary heart disease

Scandinavian Journal of Clinical Laboratory Investigation (2002) 62: 59-68

31 Blaas H-G, Eriksson AG, Salvesen KÅ, Isaksen CV, **Christensen B**, Møllerløyken G, Eik-Nes S
Brains and faces in holoprocencephaly: pre- and postnatal description of 30 cases

Ultrasound in Obstetrics and Gynecology (2002) 19(1): 24-38

32 Ernest S, **Christensen B**, Gilfix BM, Mamer OA, Hosack A, Rodier M, Colmenares C, McGrath J, Bale A, Balling R, Sankoff D, Rosenblatt DS, Nadeau JH

Genetic and molecular control of folate-homocysteine metabolism in mutant mice

Mammalian Genome (2002) 13(5): 259-267

33 Mosdøl A, **Christensen B**, Retterstol L, Thelle DS

Induced changes in the consumption of coffee alter ad libitum dietary intake and physical activity level

British Journal of Nutrition (2002) 87(3)

34 Seljeflot I, Hurlen M, **Christensen B**, Fjeldly M, Klæboe LG, Arnesen H

Homocysteine as predictor of clinical events in patients with previous myocardial infarction

In: Proceedings for Atherosclerosis: Risk factors, diagnosis, and treatment Salzburg (Austria) July 7-10, 2002. Editors: Kostner GM, Kostner KM, Monduzzi Editore S.p.A.-MEDIMOND Inc. International Proceedings Division Pp. 197-201.

35 Arbour L, **Christensen B**, Delormier T, Platt R, Gilfix B, Forbes PI, Morel J, Rozen R
Spina bifida, folate metabolism, and dietary folate intake in Northern Canadian aboriginal population

International Journal of Circumpolar Health (2002) 61(4): 341-351

- 36 Retterstol L, **Paus B**, Bohn M, Bakken A, Erikssen J, Malinow MR, Berg K
Plasma total homocysteine levels and prognosis in patients with previous premature myocardial infarction: A 10-year follow-up study
Journal of Internal Medicine (2003) 253: 284-292
- 37 Thelle DS, **Paus B**
Coffee and stillbirths, via the effect of coffee on total homocysteine?
British Medical Journal (2003) February 26 Rapid response published on the Internet
- 38 Tjeldhorn L, Rand-Hendriksen S, Gervin K, Brandal K, Inderhaug E, Geiran O, **Paus B**
Rapid and efficient FBN1 mutation detection using automated sample preparation and direct sequencing as the primary strategy
Genetic Testing (2006) 10: 258-264
- 39 Rand-Hendriksen S, Tjeldhorn L, Lundby R, Semb SO, Offstad J, Andersen K, Geiran O, **Paus B**
Search for correlations between *FBN1* genotype and complete Ghent phenotype in 44 unrelated Norwegian patients with Marfan syndrome.
American Journal of Medical Genetics Part A (2007) 143A:1968-1977
- 40 Rand-Hendriksen S, Lundby R, Tjeldhorn L, Andersen K, Offstad J, Semb SO, Smith HJ, **Paus B**, Geiran O
Prevalence data on all Ghent features in a cross-sectional study of 87 adults with proven Marfan syndrome
European Journal of Human Genetics (2009) 17(10): 1222-1230
- 41 Lundby R, Rand-Hendriksen S, Hald JK, Lilleås FG, Pripp AH, Skaar S, **Paus B**, Geiran O, Smith HJ.
Dural ectasia in Marfan syndrome: A case control study
American Journal of Neuroradiology (2009) 30(8): 1530-1540
- 42 Nordstrøm M, Hansen BH, **Paus B**, Kolset SO.
Accelerometer-determined physical activity and walking capacity in persons with Down syndrome, Williams syndrome and Prader-Willi syndrome
Res Dev Disabil. 2013; 34: 4395-403.
- 43 Drolsum L, Rand-Hendriksen S, **Paus B**, Lundby R, Geiran O, Semb S-O.
Ocular findings in 87 adults with Ghent-1 verified Marfan syndrome
Acta Ophthalmologica 2015 93(1):46-53.
- 44 Nordstrøm M, **Paus B**, Andersen LF, Kolset SO.
Dietary aspects related to health and obesity in Williams syndrome, Down syndrome and Prader-Willi syndrome
Food and Nutrition Research 2015 59:25487 doi:10.3402/fnr.v59.25487eCollection 2015.
- 45 Abrahamsen BJ, Kulseth M-A, **Paus B**.

A nineteen year old male with relapsing bilateral pneumothorax, hemoptysis and intrapulmonary cavitory lesions, diagnosed with Ehlers-Danlos' syndrome and a novel missense mutation in COL3A1
CHEST 2015 147(5):e166-e170.

46 Tjeldhorn L, Amundsen SS, Barøy T, Rand-Hendriksen L, Geiran O, Frengen E, **Paus B**. Qualitative and quantitative analysis of FBN1 mRNA from 16 patients with Marfan syndrome
BMC Medical Genetics 2015 16:113 DOI 10.1186/s12881-015-0260-4

47 Nordstrøm M, Retterstøl K, **Paus B**, Kolset SO. The prevalence of metabolic risk factors of atherosclerotic cardiovascular disease in Williams syndrome, Prader-Willi syndrome, and Down syndrome
Journal of Intellectual & Developmental Disability 2016
DOI:10.3109/13668250.2016.1167845

48 Krohg-Sørensen K, Lingaas S, Lundblad R, Seen E, **Paus B**, Geiran OR. Cardiovascular surgery in Loeys-Dietz syndrome types 1-4
European Journal of Cardio-Thoracic Surgery 2017 0: 1-7 DOI: 10.1093/ezx147

49 Henriksen MW, Breck H, von Tetzchner S, **Paus B**, Skjeldal OH, Brodtkorb E. Epilepsy in classic Rett syndrome: Course and characteristics in adult age.
Epilepsy Res. 2018 Sep;145:134-139. doi: 10.1016/j.epilepsyres.2018.06.012. Epub 2018 Jun 23.

50 Henriksen MW, Ravn K, **Paus B**, von Tetzchner S, Skjeldal OH. De novo mutations in SCN1A are associated with classic Rett syndrome: a case report. *BMC Med Genet.* 2018 Oct 11;19(1):184. doi: 10.1186/s12881-018-0700-z. PMID: 30305042

51 Riise N, Lindberg BR, Kulseth MA, Fredwall SO, Lundby R, Estensen ME, Drolsum L, Merckoll E, Krohg-Sørensen K, **Paus B**. Clinical diagnosis of Larsen syndrome, Stickler syndrome and Loeys-Dietz syndrome in a 19-year old male: a case report. *BMC Med Genet.* 2018 Aug 31;19(1):155. doi: 10.1186/s12881-018-0671-0. PMID:30170566

52 Vanem TT, Geiran OR, Krohg-Sørensen K, Røe C, **Paus B**, Rand-Hendriksen S. Survival, causes of death, and cardiovascular events in patients with Marfan syndrome. *Mol Genet Genomic Med.* 2018 Nov;6(6):1114-1123. doi: 10.1002/mgg3.489. Epub 2018 Nov 4. PMID: 30393980

53 Bøker T, Vanem TT, Pripp AH, Rand-Hendriksen S, **Paus B**, Smith HJ, Lundby R. Dural ectasia in Marfan syndrome. A 10-year follow-up study. *Spine J* 2019;19(8): 1412-1421.

54 Bratlie S, Halvorsen K, Myskja BK, Mellegård H, Bjorvatn C, Frost P, Heiene G, Hoffmann B, Holst-Jensen A, Holst-Larsen T, Malnes R, **Paus B**, Sandvig B, Sjøli S,

Skarstein B, Thorseth MB, Våge DI, Borge OJ. A novel governance framework for GMO: A tiered, more flexible regulation for GMOs would help to stimulate innovation and public debate. *EMBO Rep. Journal* 2019 May 20(5) pii: e47812 doi:p.152252/embr.201947812 EpubApr 23

55 Henriksen MW, Breck H, von Tetzchner S, **Paus B**, Skjeldal OH. Medical issues in adults with Rett syndrome – a national survey. *Developmental Neurorehabilitation* 2019; 25: 1-7. doi:10.1080/17518423.2019.1646341

56 Pope MK, Ratajska A, Johnsen H, Rypdal KB, Sejersted Y, **Paus B**. Diagnostics of hereditary connective tissue disorders by genetic next generation sequencing. *Genetic Testing and Molecular Biomarkers* 2019; 23: 11. <https://doi.org/10.1089/gtmb.2019.0064>

57 Vanem T-T, Bøker T, Sandvik GF, Kirkhus E, Smith H-J, Lundby R, Andersen K, Krohg-Sørensen K, Røe C, **Paus B**, Geiran OG, Drolsum L, Rand-Hendriksen S. Marfan syndrome: Further insights into evolving features from a 10-year follow-up study. *American Journal of Medical Genetics Part A* 2020; 182A: 397-408.

58 Henriksen MW, von Tetzchner S, **Paus B**, Skjeldal OH. Genetic and clinical variations in a Norwegian sample diagnosed with Rett syndrome. *Brain and Development* 2020; 42: 484-495.

59. Ratajska A, Vigeland M, Wirgenes KV, Krohg-Sørensen K, **Paus B**. The use of segregation analysis in interpretation of sequence variants in *SMAD3*. A case report. *Mol Genet Genomic Med.* 2023 Feb;11(2):e2107. doi: 10.1002/mgg3.2107. Epub 2022 Dec

60 Seim BE, Holt MF, Ratajska A, Michelsen A, Ringseth MM, Halvorsen BE, Skjelland M, Kvitting J-P E, Lundblad R, Krohg-Sørensen K, Osnes LTN, Aukrust P, **Paus B**, Ueland T (shared last authorship). Markers of extracellular matrix remodeling and systemic inflammation in patients with heritable thoracic aortic diseases. *Front Cardiovasc Med.* 2022 Dec 20;9:1073069. doi: 10.3389/fcvm.2022.1073069. eCollection 2022. *In press* 24.11.22

61 Iversen N, Henriksson CE, Sletten M, Le MS, Lindberg BR, Andersen R, **Paus B**. Heterozygosity for the Budapest 3 mutation in *SERPINC1* in a family with thrombophilia and structural anomalies of the inferior vena cava. *Thromb J.* 2024 Aug 12;22(1):75. doi: 10.1186/s12959-024-00644-1.PMID: 39129027

62. Bøker T, Vanem T-T, Rand-Hendriksen S, **Paus B**, Smith HJ, Lundby R. The natural history of protrusio acetabuli in Marfan syndrome and other hereditary connective tissue disorders. A 10-year follow-up CT study. *Orphanet Journal of Rare Diseases* 2025 *In press* 24.02.25

63. Seim BE,

64 Sekkelsten A,

Book chapters

65 Solberg CO, Digranes A, Haram KO, Bratland SZ, Markestad TJ, **Christensen B**
Infeksjoner i svangerskapet
DMB&B MEDICUS Oslo (1993), ISBN 82-91396-01-9

66 **Christensen B**, Rosenblatt DS
Effects of folate deficiency on embryonic development
In: Bailliere's Clinical Haematology. International practice and research: Megaloblastic Anaemias. Ed.: Wickramasinghe S. Bailliere Tindall. London (1995) 8(3):617-637

67 **Christensen B**, Rosenblatt DS
The role of folate and vitamin B12 in the central nervous system: Lessons from inborn errors
In: Epilepsy and other neurological disorders in coeliac disease Eds.: Gobbi, G and Banchini G. John Libbey & Company Ltd Medical Books, London (1997) p39-45

68 **Paus B**, Undlien D
Indremedisin
Genetikk ved indremedisinske sykdommer
*Forlaget Vett og Viten (2017) Chapter 5 p
Revised (2nd ed.) (2021)*

Book

69 **Paus B**: Klinisk genetikk. En innføringsbok ISBN 978-82-05-39343-1
Gyldendal Akademisk Oslo (2009) 245s

Other publications

70 Hexeberg E, Schneede J, Øgreid D, Rødahl E, Skulstad S, and **Christensen B**
Årsmøte i leger i vitenskapelige stillinger (LVS)

The Journal of the Norwegian Medical Association (1992) 112(4):560-1

71 Øgreid D, **Christensen B**, Collett K
Stipendiatenes valg av yrkesforening 1993

The Journal of the Norwegian Medical Association (1992) 112(30):3904-5

72 Rand-Hendriksen S, **Christensen B**.

Magnettomografi av sentralnervesystemet (Korrespondanse)

The Journal of the Norwegian Medical Association (1999)119(2):264

73 **Christensen B**.

Om genteknologi og etikk. (Bokanmeldelse)

The Journal of the Norwegian Medical Association (2000)120(6): 722

74 **Christensen B**.

Ambisiøst og uklart om biologisk medisin. (Bokanmeldelse)

The Journal of the Norwegian Medical Association (2001)

75 **Christensen B**, Mosdol A, Thelle DS Abstention from filtered coffee reduces the levels of homocysteine and cholesterol. Reply. (Korrespondanse)

The American Journal of Clinical Nutrition (2002) 75

76 **Paus B**

Pedagogisk og innholdsrik genetikkbok

Tidsskr Nor Legeforen 2013; 133: 1633

77 **Paus B**

Grundig ”crash-kurs” i cellebiologi med mer

Tidssk Nor Legeforen 2013; 133: 2069

78 **Paus B**

Når vi snakker om gener

The Journal of the Norwegian Medical Association (2015); 135(8): 774-5.

79 **Paus B**

Kanskje teste, ofte utrede, alltid veilede ([Perhaps test, often explore, always counsel](#)). Tidsskr Nor Laegeforen. 2018 Sep 3;138(13). doi: 10.4045/tidsskr.18.0574. Print 2018 Sep 4. English, Norwegian. No abstract available. PMID:30180484

80 **Paus B** B Paus responds Re: Kanskje teste, ofte utrede, alltid veilede ([Perhaps test, often explore, always counsel](#)).

The Journal of the Norwegian Medical Association (2018); Oct 15 138(16) doi: 10.4045/tidsskr.18.0735

81 Gjervik P, **Paus B**

Mutasjoner som endrer proteinets funksjon

The Journal of the Norwegian Medical Association (2021); 141: 1214-15.

82 **Paus B**

Grundig om fosterdiagnostikk (Bokanmeldelse)

The Journal of the Norwegian Medical Association (2001); 141: 1217.