Publikationsverzeichnis

A) Originalarbeiten (IF = *impact factor* nach ISI)

1. **Reiss, J.** , Kleinhofs, A., Klingmüller, W. (1987)
   Cloning of seven differently complementing DNA fragments with *chl* functions from *Escherichia coli* K12
   Mol Gen Genet 206: 352-355 (IF = 3)
2. **Reiss, J**., Klingmüller, W. (1987)
   Direct selection of recombinant plasmids with chlorate
   FEMS Microbiol Lett 43: 201-205 (IF = 2)
3. Krawczak, M., **Reiss, J**., Schmidtke, J., Rössler, U. (1989)
   Polymerase chain reaction: replication errors and reliability of gene diagnosis
   Nucleic Acids Res 17: 2197-2201 (IF = 5)
4. Wagner, M., **Reiss, J.**, Hentemann, M., Thies, U. (1989)
   *Msp*I RFLP for Duchenne muscular dystrophy cDNA subclone 9
   Nucleic Acids Res 17:3328 (IF = 5)
5. **Reiss, J**., Krawczak, M., Gal, A., Zerres, K., Kaiser, R., Weber, J. (1989)
   Risikoschwangerschaft einer Anlageträgerin für Cystische Fibrose (Mukoviszidose) mit einem neuen Partner
   Monatsschr Kinderheilk 137:451-453 (IF = 0.25)
6. Slomski, R., **Reiss, J**., Jungermann, M. (1989)
   Application of non-radioactive methods of DNA detection in analysis of human genetic disorders
   Acta Biochim Pol 36:311-321 (IF = 1)
7. **Reiss, J**., Neufeldt, U., Wieland, K., Zoll, B. (1990)
   Diagnosis of Hemophilia B using the Polymerase Chain Reaction
   Blut 60:31-36 (IF = 0.25)
8. Hentemann, M., **Reiss, J**., Wagner, M., Cooper, D.N. (1990)
   Rapid detection of deletions in the *DMD* gene by amplification of deletion-prone exon sequences
   Hum Genet 84:228-232 (IF = 3)
9. **Reiss, J**., Krawczak, M., Schlösser, M., Wagner, M., Cooper, D.N. (1990)
   The effect of replication errors on the mismatch analysis of PCR amplified DNA
   Nucleic Acids Res 18:973-978 (IF = 5)
10. Wagner, M., Diedrich, U., Poszar, C., Becker, P.E., **Reiss, J**. (1990)
    Zur Differentialdiagnostik gutartig verlaufender Muskeldystrophien durch molekulargenetische Methoden
    Nervenarzt 61:244-247 (IF = 1)
11. Wagner, M., Schlösser, M., **Reiss, J.** (1990)
    Direct gene diagnosis of cystic fibrosis by allel specific polymerase chain reactions
    Mol Biol Med 7:359-364 (IF = 1)
12. **Reiss, J.,** Schlösser, M., Wagner, M., Lenz, U., Krawczak, M., Amman, G., Klösser, S., Böwing, B. (1990)
    Direkte Gendiagnostik bei Cystischer Fibrose
    Monatsschr Kinderheilkde 138:434-437 (IF = 0.25)

13. European Working Group on CF Genetics (1990)
    Gradient of distribution in Europe of the major CF mutation and of its associated halotype
    Hum Genet 86:436-445 (IF = 3)

14. Reis, A., Bremer, S., Schlösser, M., Dück, M., Böhm, I.,. Hundrieser, J., Macek, M., Stuhrmann, M., Wagner, M., Dörk, T., Schnieders, F., Posselt, H.G., Wahn, U., **Reiss, J.**, Trefz, F., Tümmler, B., Krawczak, M., Schmidtke, J. (1990)
    Distribution pattern of the delta F508 mutation in the *CFTR* gene on CF-linked marker haplotypes in the German population
    Hum Genet 85:421-422 (IF = 3)

15. Hundrieser, J., Bremer, S., Peinemann, F., Stuhrmann, M., Hoffknecht, N., Wulf, B., Schmidtke, J., **Reiss, J.**, Maaß, G., Tümmler, B. (1990)
    Frequency of the F508 deletion in the *CFTR* gene in Turkish cystic fibrosis patients
    Hum Genet 85:409-410 (IF = 3)

16. Worldwide survey of the ΔF508 mutation – report from the Cystic Fibrosis genetic analysis consortium (1990)
    Am J Hum Gen 47:354-359 (IF = 11)

17. Berg, L.P., Wieland, K., Millar, D.S., Schlösser, M., Wagner, M., Kakkar, V.V., **Reiss, J**., Cooper, D.N. (1990)
    Detection of a novel point mutation causing haemophilia A by PCR/direct sequencing of ectopically-transcribed Factor VIII mRNA
    Hum Genet 85:655-658 (IF = 3)

18. Pattinson, J.K., Millar, D.S., Grundy, C.B., Wieland, K., Mibashan, R.S., Martinowitz, U., McVey, J., Tan-Un, K., Videaud, M., Goosens, M., Sampietro, M., Camerino, G., Krawczak, M., **Reiss, J**., Zoll, B., Whitmore, D., Bradshaw, A., Wensley, R., Ajani, A., Mitchell, V., Rizza, C., Maia, R., Winter, P., Mayne, E.E., Kakkar, V.V., Tuddenham, E.G.D., Cooper, D.N. (1990)
    The molecular genetic analysis of haemophilia A; a directed search strategy for the detection of point mutations in the human factor VIII gene
    Blood, 76:2242-2248 (IF = 9)

19. Schloesser, M., Slomski, R., Wagner, M., Berg, L.P., Kakkar, V.V., Cooper, D.N., **Reiss, J.** (1990)
    Characterization of pathological dystrophin transcripts from the lymphocytes of a muscular dystrophy carrier
    Mol Biol Med 7:519-523 (IF = 1)

20. Peinemann, F., Wagner, M., Franke, U., Kulle, M., **Reiss, J.** (1991)
    Prenatal deletion detection in a sporadic case of Duchenne muscular dystrophy without genotype information from the affected individual
    Eur J Ped 150:256-258 (IF = 1)

21. Bal, J., Maciejko, B., Mazurczak, T., Potocka, A., Krawczak, M., **Reiss, J**. (1991)
    Frequency of the cystic fibrosis mutation ΔF508 in Poland
    Hum Genet 86:329 (IF = 3)

22. Slomski, R., Schlösser, M., Chlebowska, H., **Reiss, J.**, Engel, W. (1991)
    Detection of human spermatid-specific transcripts in the lymphocytes of males and females
    Hum Genet 87:307-310 (IF = 3)

23. **Reiss, J**., Cooper, D.N., Bal, J., Slomski, R., Cutting, G.R., Krawczak, M. (1991)
Discrimination between recurrent mutation and identity by descent: application to point mutations in exon 11 of the *CFTR* gene
Hum Genet 87:457-461 (IF = 3)

24. Bal, J., Stuhrmann, M., Schlösser, M., Schmidtke, J., **Reiss, J.** (1991)
A cystic fibrosis patient homozygous for the nonsense mutation R553X
J Med Genet 28:715-717 (IF = 5)

25. Schloesser, M., Arleth, S., Lenz, U., Bertele, R.M., **Reiss, J**. (1991)
A cystic fibrosis patient with the nonsense mutation G542X and the splice site mutation 1717-1
J Med Gen 28:878-880 (IF = 5)

26. Plieth, J., Rininsland, F., Schloesser, M., Cooper, D.N., **Reiss, J**. (1992)
Single strand conformation polymorphism (SSCP) analysis of the *CFTR* gene reliably detects more than one third of non-ΔF508 mutations in German Cystic Fibrosis patients
Hum Genet 88:283-287 (IF = 3)

27. Bal, J., Rininsland, F., Osborne, L., **Reiss, J**. (1992)
Simple non-radioactive detection of the *CFTR* mutation N1303K by artificial creation of a restriction site
Mol Cell Probes 6:9-11 (IF = 2)

28. Berg, L.P., Grundy, C.B., Thomas, F., Millar, D.S., Green, P.J., Slomski, R., **Reiss, J**., Kakkar, V.V., Cooper, D.N. (1992)
De novo splice site mutation in the antithrombin III (*AT3*) gene causing recurrent thrombosis: Demonstration of exon skipping by ectopic transcript analysis
Genomics 13:1359-1361 (IF = 3)

29. Rininsland, F., Hahn, A., Niemann-Seyde, S., Slomski, R., Hanefeld, F., **Reiss, J**. (1992)
Identification of a new *DMD* gene deletion by ectopic transcript analysis
J Med Genet 29:647-651 (IF = 5)

30. Slomski, R., Schloesser, M., Berg, L.P., Wagner, M., Kakkar, V.V., Cooper, D.N., **Reiss, J.** (1992)
Omission of exon 12 in CFTR gene transcripts
Hum Genet 89:615-619 (IF = 3)

31. Slomski, R., Braulke, I., Behrend, C., Schröder, E., Colombo, J.P., **Reiss, J.** (1992)
Ornithine transcarbamylase deficiency (*OTC*) in a female patient with a de novo deletion of the paternal X chromosome
Hum Genet 89:632-634 (IF = 3)

32. Osborne, L., Santis, G., Schwarz, M., Klinger, K., Dörk, T., McIntosh, I., Schwartz, M., Nunes, V., Macek, M.Jr., **Reiss, J**., Highsmith, W.E.Jr., McAhon, R., Novelli, G., Malik, N., Bürger, J., Anvret, M., Wallace, A., Williams, C., Mathew, C., Rozen, R:, Graham, C., Gasparini, P., Bal, J., Cassiman, J., Balassopoulou, A., Davidow, L., Raskin, S., Kalaydjieva, L., Kerem, B., Richard, S., Simon-Bouy, B., Super, M., Wulbrand, U., Keston, M., Estivill, X., Vavrova, V., Friedman, K.J., Barton, D., Dallapiccola, B., Stuhrmann, M., Beards, F., Hill, A.J.M., Pignatti, P., Cuppens, H., Angelicheva, D., Tümmler, B., Brock, D.J.H., Casals, T., Macek, M., Schmidtke, J., Magee, A.C., Bonizzato, A., DeBoeck, C., Kuffardjieva, A., Hodson, M., Knight, R.A. (1992)
Incidence and expression of the N1303K mutation of the cystic fibrosis (*CFTR*) gene
Hum Genet 89:653-658 (IF = 3)

33. Krawczak, M., **Reiss, J.**, Cooper, D.N. (1992)
The mutational spectrum of single base-pair substitutions in mRNA splice junctions of human genes: Causes and consequences
Hum Genet 90:41-54 (IF = 3)

34. Niemann-Seyde, S., Slomski, R., Rininsland, F., Ellermeyer, U., Kwiatkowska, J., **Reiss, J.** (1992)
Molecular genetic analysis of 67 patients with Duchenne/Becker muscular dystrophy
Hum Genet 90:65-70 (IF = 3)

35. Bal, J., Mazurczak, T., **Reiss, J**. (1992)
The frequency of mutations in exon 11 of the *CFTR* gene in Polish cystic fibrosis patients
Acta Biochim Pol 39:245-249 (IF = 1)

36. **Reiss, J**., Lenz, U., Rininsland, F., Ballhausen, P., Drews, D., Posselt, H.G. (1992)
A novel *CFTR* mutation , 4053delA, detected by non-radioactive SSCP analysis
Hum Genet 90:303-304 (IF = 3)

37. **Reiss, J**., Ellermeyer, U., Schloesser, M., Fuhrmann, W., Drews, D., Posselt, H.G. (1993)
Two cystic fibrosis patients with the genotype G542X/G551D
Hum Genet 91:78-79 (IF = 3)

38. Will, K., **Reiss, J**., Dean, M., Schlösser, M., Slomski, R., Schmidtke, J., Stuhrmann, M. (1993)
CFTR transcripts are undetectable in lymphocytes and respiratory epithelial cells of CF patients homozygous for the nonsense mutation R553X
J Med Genet 30:833-837 (IF = 5)

39. **Reiss, J.**, Ellermeyer, U., Rininsland, F., Ballhausen, P., Lenz, U., Wagner, S., Schlösser, M. (1993)
A comprehensive *CFTR* mutation analysis of German CF patients
Hum Mol Genet 2:809-811 (IF = 9)

40. **Reiss, J**., Rininsland, F. (1994)
An explanation for the constitutive exon 9 cassette splicing of the *DMD* gene
Hum Mol Genet 3:295-298 (IF = 9)

41. Rininsland, F., **Reiss, J**. (1994)
Microlesions and polymorphisms in the Duchenne/Becker muscular dystrophy gene
Hum Genet 94:111-116 (IF = 3)

42. Rickers, A., Rininsland, F., Osborne, L., **Reiss, J.** (1994)
Skipping of multiple *CFTR* exons is not a result of single exon omissions
Hum Genet 94:311-313 (IF = 3)

43. Seemann, P., Stoppe, G., Laccone, F., **Reiss, J.** (1995)
Absence of mutations in the Apolipoprotein E (*APOE*) gene of patients with Alzheimer disease
Hum Mut 5:103-104 (IF = 6)

44. Shalata, A., Mandel, H., **Reiss, J**., Szargel, R., Cohen-Akenine, A., Dorche, C., Zabot; M.T., Van Gennip, A., Abeling, N., Berant, M., Cohen, N. (1998)
Localization of a gene for molybdenum cofactor deficiency, on the short arm of chromosome 6, by homozygosity mapping
Am J Hum Genet 63:148-154 (IF = 11)

45. **Reiss, J**., Cohen, N., Dorche, C., Mandel, H., Mendel, R.R., Stallmeyer, B., Zabot, M.T., Dierks, T. (1998)
Mutations in a polycistronic nuclear gene associated with molybdenum cofactor deficiency
Nature Genetics 20:51-53 (IF = 31)

46. **Reiss, J**., Christensen, E., Kurlemann, G., Zabot, M.T., Dorche, C. (1998)
Genomic structure and mutational spectrum of the bicistronic *MOCS1* gene defective in molybdenum cofactor deficiency type A
Hum Genet 103:639-644 (IF = 3)

47. Stallmeyer, B., Schulze, J., Schwarz, G., Nerlich, A., **Reiss, J**., Kirsch, J., Mendel, R.R. (1999)
The neurotransmitter receptor-anchoring protein gephyrin reconstitutes molybdenum cofactor biosynthesis in bacteria, plants, and mammalian cells
Proc Natl Acad Sci USA 96:1333-1338 (IF = 11)

48. Stallmeyer, B., Drugeon, G., **Reiss, J.**, Haenni, A.L., Mendel, R.R. (1999)
Human molybdopterin synthase gene:
Identification of a bicistronic transcript with overlapping reading frames
Am J Hum Genet 64:698-705 (IF = 11)

49. **Reiss, J**., Cohen, N., Stallmeyer, B., Mendel, R.R., Dorche, C. (1999)
Human molybdopterin synthase gene:
Genomic structure and mutations in molybdenum cofactor deficiency type B
Am J Hum Genet 64:706-711 (IF = 11)

50. **Reiss, J**., Christensen, E., Dorche, C. (1999)
Molybdenum cofactor deficiency: First prenatal genetic analysis
Prenat Diagn 19:386-388 (IF = 2)

51. Shalata, A., Mandel, H., Dorche, C., Zabot, M.T., Shalev, S., Hugeirat, Y., Arieh, D., Ronit, Z., **Reiss, J.**, Anbinder, Y., Cohen, N. (2000)
Prenatal diagnosis and carrier detection for molybdenum cofactor deficiency type A in Northern Israel using polymorphic DNA markers
Prenat Diagn 20:7-11 (IF = 2)

52. Finckh, U., von der Kammer, H., Velden, J., Michel, T., Andresen, B., Deng, A., Müller-Thomsen, T., Zuchowski, K., Menzer, G., Mann, U., Papassotiropoulos, A., Heun, R., Zurdel, J., Holst, F., Benussi, L., Stoppe, G., **Reiss, J.**, Miserez, A.R., Staehelin, H.B., Rebeck, G.W., Hyman, B.T., Binetti, G., Hock, C., Growdon, J.H., Nitsch, R.M. (2000)
Genetic association of the Cystatin C gene with late-onset Alzheimer disease
Arch Neurol 57:1579-1583 (IF = 4)

53. **Reiss, J.**, Gross-Hardt, S., Christensen, E., Schmidt, P., Mendel, R.R., Schwarz, G. (2001)
A mutation in the gene for the neurotransmitter receptor-clustering protein *Gephyrin* causes a novel form of molybdenum cofactor deficiency
Am J Hum Genet 68:208-213 (IF = 11)

54. Menzer, G., Müller-Thomsen, T., Meins, W., Alberici, A., Binetti, G., Hock, C., Nitsch, R.M., Stoppe, G., **Reiss, J.,** Finckh, U. (2001)
Non-replication of association between Cathepsin D genotype and late onset Alzheimer disease
Am J Med Genet 105:179-182 (IF = 2)

55. Gross-Hardt, S., **Reiss, J.** (2002)
The bicistronic *MOCS*1 gene has alternative start codons on two mutually exclusive exons
Mol Genet Metab 76: 340-3 (IF = 2)

56. Lee, H.J., Adham, I.M., Schwarz, G., Kneussel, M., Sass, J.O., Engel, W., **Reiss, J.** (2002)
Molybdenum cofactor-deficient mice resemble the phenotype of human patients
Hum Mol Genet 11:3309-3317 (IF = 9)

57. **Reiss, J.**, Johnson, J. (2003)
Mutations in the molybdenum cofactor bioynthesis genes *MOCS*1, *MOCS*2 and *GEPH*
Hum Mutat 21:569-576 (IF = 6)

58. Sass, J.O., Kishikawa, M., Puttinger, R., **Reiss, J.**, Erwa, W., Shimizu, A., Sperl, W. (2003) Hypohomocysteinemia and highly increased proportion of S-sulfonated plasma transthyretin in molybdenum cofactor deficiency
J Inherit Metab Dis 26: 80-82 (IF = 2)

59. Finckh, U., von Hadel, K., Müller-Thomsen, T., Alberici, A., Binetti, G., Hock, C., Nitsch, R.M., Stoppe, G., **Reiss, J.**, Gal, A. (2003)
Association of Alzheimer disease with a genotype of PLAU, the gene encoding urokinase-type plasminogen activator located on chromosome 10q22.2
Neurogenetics 4: 213-217 (IF = 3)

60. Schwarz, G., Araujo, J.A.S., Haenzelmann, P., Mendel, R.R., Lee, H.J., Adham, I.M., Engel, W., **Reiss, J.** (2004)
Rescue of the lethal molybdenum cofactor deficiency by a biosynthetic precursor from *Escherichia coli*
Hum Mol Genet 13: 1249-1255 (IF = 9)

61. Günther, K., von Hadeln, K., Müller-Thomsen, T., Alberici, A., Binetti, G., Hock, C., Nitsch, R.M., Stoppe, G., **Reiss, J.**, Gal, A., Finckh, U. (2004)
Possible association of mitochondrial transcription factor A (TFAM) genotype with sporadic Alzheimer disease
Neurosci Lett 369: 219-223 (IF = 2)

62. **Reiss, J.**, Bonin, M., Schwegler, H., Sass, J.O., Garrattini, E., Wagner, S., Lee, H.J., Engel, W., Riess, O., Schwarz, G. (2005)
The pathogenesis of molybdenum cofactor, its delay by maternal clearance and its expression pattern in microarray analysis
Mol Genet Metab 85: 12-20 (IF = 2)

63. Leimkühler, S., Charcosset, M., Philippe Latour, P., Dorche, C., Kleppe, S., Scaglia, F., Szymczak, I., Schupp, P., Hahnewald, R., **Reiss, J.** (2005)

    Ten novel mutations in the Molybdenum Cofactor Genes MOCS1 and MOCS2 and in vitro characterization of a MOCS2 mutation that abolishes the binding ability of molybdopterin synthase
    Hum Genet, im Druck (IF = 3)

Kumulativer *impact factor* der akzeptierten Originalarbeiten: 287

B) Übersichtsartikel

1. **Reiss, J.,** Cooper, D.N. (1990)
   Application of the polymerase chain reaction to the diagnosis of human genetic disease
   Hum Genet 85:1-8
2. Peinemann, F., **Reiss, J**. (1991)
   Grundlagen der DNA-Diagnostik am Beispiel der Cystischen Fibrose
   Monatsschr Kinderheilkde 139:83-88
3. **Reiss, J**. (1991)
   Invited review: The polymerase chain reaction and its potential role in clinical diagnostics and research
   J Intern Med 230:391-395
4. Cooper, D.N., Berg, L.P., Kakkar, V.V., **Reiss, J.** (1994)
   Ectopic (Illegitimate) Transcription: New posibilities for the analysis and diagnosis of human genetic disease
   Ann Med 26:9-14
5. **Reiss, J.** (2000)
   Genetics of molybdenum cofactor deficiency
   Hum Genet 106:157-163

C) Buchbeiträge

1. Stuhrmann, M., Dörk, T., Krawczak, M., Dueck, M., Banholzer, U., Domagk, J., Hoffknecht, N., Posselt, H.G., Reis, A., Schlösser, M., Trefz, F., Wagner, M., Wahn, U., Wulf, B., Schmidtke, J., **Reiss, J**., Tümmler, B.
   Genotype/phenotype correlations in cystic fibrosis patients
   In: Tsui, L.C., et al. (1991)
   The identification of the CF (Cystic Fibrosis) gene
   Plenum Press, New York, 97-103
2. **Reiss, J**.
   Ectopic transcription in the analysis of human genetic disease
   In: Rolfs, A., Schuller, U., Finckh, U., Weber-Rolfs, I., (1992)
   PCR: Clinical diagnostics and research
   Springer Verlag, Berlin, A63-A66
3. Wagner, M., **Reiss, J.**
   The cystic fibrosis gene
   In: Verma, R.S. (1993)
   Advances in Genome Biology II
   JAI Press, Greenwich, 69-98
4. **Reiss, J.,** Anke, M.
   Molybdän und Cobalt
   In: Biesalski, K., Köhrle, J., Schümann, J. (2002)
   Vitamine, Spurenelemente und Mineralstoffe
   Georg Thieme Verlag, Stuttgart; New York, 218-221

D) Sonstige Veröffentlichungen

Reiss, J, Wieland, K., Zoll, B. (1988)
Fortschritte in der DNA-Analyse mit Hilfe der PCR-Methode am Beispiel der Hämophilie B
BioEngineering 4/88:70-74

Zoll, B., **Reiss, J.** (1989)
Die molekulargenetische Diagnose der Hämophilien A und B
Niedersächsisches Ärzteblatt 6/1989:16-21

Reiss, J, Zoll, B. (1989)
Hämophilie-Diagnostik durch DNA-Amplifizierung
Niedersächsisches Ärzteblatt 8/1989

Hentemann, M., Wagner, M., **Reiss, J.** (1989)
Detection of deletions in DMD patients by amplification of exon sequences
Cytogenet Cell Genet51:1013

Wagner, M., Hentemann, M., **Reiss, J.** (1989)
A new intragenic RFLP in the *DMD* gene detectable by cDNA hybridization
Cytogenet Cell Genet 51:1100

Korabecna, M., Peinemann, F., **Reiss, J.** (1990)
Direct carrier detection for female relatives of DMD patients
In: Macek, M., Ferguson-Smith, M.A., Spala, M. (eds)
Early fetal diagnosis: Recent progress and public health impact.
Karolinum – Charles University Press, Prague

Peinemann, F., **Reiss, J**. (1991)
DNA-Diagnostik am Beispiel der Cystischen Fibrose
TW Gynäkologie 4:29-40

Reiss, J., Krawczak, M., Wagner, M., Engel, W., Schmidtke, J. (1991)
CF-Risiko-Informationen nicht vorenthalten
Med Genetik 1/1991:61

Reiss, J. (1993)
Would you sign your criticism?
Lancet 341:1097

Stuhrmann-Spangenberg, M., Auhlela-Scholz, C., Dworniczak, B., **Reiss, J.** (1997)
Leitlinien zur Molekulargenetischen Diagnostik der Cystischen Fibrose
Med Genetik 4:553-559

Reiss, J. (1998)
Book review: Genetic engineering with PCR: R.M. Horton and R.C. Tait (Editors)
Hum Genet 102:706