

Morak M, Schaefer K, Steinke-Lange V, et al. **Full-length transcript amplification and sequencing as universal method to test mRNA integrity and biallelic expression in mismatch repair genes.** *Eur J Hum Genet.* July 2019. doi:[10.1038/s41431-019-0472-8](https://doi.org/10.1038/s41431-019-0472-8)

Neubauer K, Boeckelmann D, Koehler U, et al. **Hereditary neuralgic amyotrophy in childhood caused by duplication within the SEPT9 gene: A family study.** *Cytoskeleton (Hoboken).* 2019;76(1):131-136. doi:[10.1002/cm.21479](https://doi.org/10.1002/cm.21479)

Hallermayr A, Graf J, Koehler U, et al. **Extending the critical regions for mutations in the non-coding gene RNU4ATAC in another patient with Roifman Syndrome.** *Clin Case Rep.* 2018;6(11):2224-2228. doi:[10.1002/ccr3.1830](https://doi.org/10.1002/ccr3.1830)

Depienne C, Nava C, Keren B, et al. **Genetic and phenotypic dissection of 1q43q44 microdeletion syndrome and neurodevelopmental phenotypes associated with mutations in ZBTB18 and HNRNPU.** *Hum Genet.* 2017;136(4):463-479. doi:[10.1007/s00439-017-1772-0](https://doi.org/10.1007/s00439-017-1772-0)

Sandrock-Lang K, Bartsch I, Buechele N, et al. **Novel mutation in two brothers with Hermansky Pudlak syndrome type 3.** *Blood Cells Mol Dis.* 2017;67:75-80. doi:[10.1016/j.bcmd.2017.03.001](https://doi.org/10.1016/j.bcmd.2017.03.001)

Coci EG, Koehler U, Liehr T, et al. **CANPMR syndrome and chromosome 1p32-p31 deletion syndrome coexist in two related individuals affected by simultaneous haplo-insufficiency of CAMTA1 and NIFA genes.** *Mol Cytogenet.* 2016;9:10. doi:[10.1186/s13039-016-0219-y](https://doi.org/10.1186/s13039-016-0219-y)

Kleinle S, Koehler U, Gonzalez Fassreiner D, Holinski-Feder E. **Genetische Untersuchungen in der Reproduktionsmedizin.** *Journal für Reproduktionsmedizin und Endokrinologie* 2015 (2): 57-64. doi: <https://www.kup.at/kup/pdf/12848.pdf>

Koehler U, Pabst B, Pober B, Kozel B. **Clinical utility gene card for: Williams-Beuren Syndrome [7q11.23].** *Eur J Hum Genet.* 2014;22(9). doi:[10.1038/ejhg.2014.28](https://doi.org/10.1038/ejhg.2014.28)

Seifert B, Paulmann B, Seifert D, Brey S, Gaßner C, Kwiatkowski B, Schön U, Koehler U, Holinski-Feder E. **Studie zur klinischen Etablierung der Präimplantationsdiagnostik.** *Journal für Reproduktionsmedizin und Endokrinologie* 2014; 11(1):12-17. Doi:<https://www.kup.at/kup/pdf/12122.pdf>

Czeschik JC, Bauer P, Buiting K, et al. **X-linked intellectual disability type Nascimento is a clinically distinct, probably underdiagnosed entity.** *Orphanet J Rare Dis.* 2013;8:146. doi:[10.1186/1750-1172-8-146](https://doi.org/10.1186/1750-1172-8-146)

Koehler U., Schoen U., Mayer V., Holinski-Feder. E. **Preimplantation Genetic Diagnosis for Monogenic Disorders and Chromosomal Rearrangements – the German Perspective.** *Journal für Reproduktionsmedizin und Endokrinologie* 2013; Special Issue 1:36-42. doi:<https://www.kup.at/kup/pdf/11278.pdf>

Würfel W, Suttner R, Shakeshaft D, et al. **Pregnancy and Birth After a Two-Step PGD: Polar Body Diagnosis for Hemophilia A and Array CGH on Trophectoderm Cells for Chromosomal Aberrations.** *Geburtshilfe Frauenheilkd.* 2013;73(8):812-814. doi:[10.1055/s-0033-1350705](https://doi.org/10.1055/s-0033-1350705)

Dikow N, Maas B, Gaspar H, Kreiss-Nachtsheim M, Engels H, Kuechler A, Garbes L, Netzer C, Neuhaus TM, Koehler U, et al. **The phenotypic spectrum of duplication 5q35.2-q35.3 encompassing NSD1: Is it really a reversed sotos syndrome?** *Am J Med Genet A.* 2013 Sep;161(9):2158-66.

Gothwal M, Nakamura L, Hainmann I, et al. **Molecular genetic analysis of a patient with moderate hemophilia A and psychomotor developmental delay.** *Klin Padiatr.* 2013;225(3):175-176. doi:[10.1055/s-0033-1341490](https://doi.org/10.1055/s-0033-1341490)

Kluger G, Koehler U, Neuhaus TM, Pieper T, Staudt M, von Stülpnagel C. **Generalized epilepsy in two patients with 5p duplication.** *Neuropediatrics.* 2013 Aug;44(4):225-9.

Suttner R, Shakeshaft D, Koehler U, Schön U, Hararsim T, Wagner A, Holinski-Feder E, Rost I, Würfel W. **First experiences with PGD after trophoctoderm biopsy at Kinderwunsch Centrum Munich (KCM), Germany.** *Reprod BioMed Online* 2012; 24:48.

U. Schön, U. Koehler, V. Mayer, A. Stegerer, K. Sendelbach, M. Locher, B. Paulmann, D. Shakeshaft, R. Suttner, B. Seifert, W. Würfel, E. Holinski-Feder. **Increased pregnancy rates after trophoctoderm biopsy for PGD of monogenic diseases and chromosomal aberrations.** *Reproductive BioMedicine Online* 2012; 24:55-56.

Koehler U, Mayer V, Schön U, Stegerer A, Sendelbach K, Locher M, Paulmann B, Shakeshaft D, Suttner R, Seifert B, Würfel W, Holinski-Feder E. **Steigerung der Schwangerschaftsrate durch Präimplantationsdiagnostik für monogene Erkrankungen und chromosomale Aberrationen nach Trophoctodermibiopsie.** *Arch Gynecol Obstet* 2012; 286:49-279.

Koehler U, Schön U, Mayer V, Stegerer A, Sendelbach K, Locher M, Paulmann B, Shakeshaft D, Suttner R, Seifert B, Würfel W, Holinski-Feder E. **Preimplantation genetic diagnosis for monogenic diseases and chromosomal aberrations after trophoctoderm biopsy- results from 2010 to 2011.** *European Journal of Human Genetics* 2012; 20:140.

Canis M, Lechner A, Mack B, et al. **CD133 induces tumour-initiating properties in HEK293 cells.** *Tumour Biol.* 2013;34(1):437-443. doi:[10.1007/s13277-012-0568-z](https://doi.org/10.1007/s13277-012-0568-z)

Schmidt B, Udink ten Cate F, Weiss M, Koehler U. **Cardiac malformation of partial trisomy 7p/monosomy 18p and partial trisomy 18p/monosomy 7p in siblings as a result of reciprocal unbalanced malsegregation--and review of the literature.** *Eur J Pediatr.* 2012;171(7):1047-1053. doi:[10.1007/s00431-012-1682-z](https://doi.org/10.1007/s00431-012-1682-z)

Canis M, Lechner A, Mack B, et al. **CD133 is a predictor of poor survival in head and neck squamous cell carcinomas.** *Cancer Biomark.* 2012;12(2):97-105. doi:[10.3233/CBM-130297](https://doi.org/10.3233/CBM-130297)

Hehr A, Paulmann P, Koehler U., Gassner C, Bals-Pratsch M, Holinski-Feder E, et al. **PID für monogene Erkrankungen nach Polkörper- und/oder Trophektodermbiopsie.** *J Reproduktionsmed Endokrinol* 2011; 8:319.

Seifert B, Schön U, Paulmann B, Seifert D, Hehr A, Koehler U, Holinski-Feder E. **Diagnostik der spinalen Muskelatrophie Typ 1 (SMA1) durch Trophektodermbiopsie von Blastozysten.** *J Reproduktionsmed Endokrinol* 2011; 8:335.

Moog U, Kutsche K, Kortum F, et al. **Phenotypic spectrum associated with CASK loss-of-function mutations.** *J Med Genet.* 2011;48(11):741-751. doi:[10.1136/jmedgenet-2011-100218](https://doi.org/10.1136/jmedgenet-2011-100218)

Morak M, Koehler U, Schackert HK, et al. **Biallelic MLH1 SNP cDNA expression or constitutional promoter methylation can hide genomic rearrangements causing Lynch syndrome.** *J Med Genet.* 2011;48(8):513-519. doi:[10.1136/jmedgenet-2011-100050](https://doi.org/10.1136/jmedgenet-2011-100050)

Koehler U, Holinski-Feder E, Ertl-Wagner B, et al. **A novel 1p31.3p32.2 deletion involving the NFIA gene detected by array CGH in a patient with macrocephaly and hypoplasia of the corpus callosum.** *Eur J Pediatr.* 2010;169(4):463-468. doi:[10.1007/s00431-009-1057-2](https://doi.org/10.1007/s00431-009-1057-2)

Koehler U., Holinski-Feder E. **Humangenetische Untersuchungen in der Pränataldiagnostik Frauenheilkunde up2date** 2010(5) 278-283.

Martin P., Koehler U. **Selbstschädigendes Verhalten bei zwei männlichen Patienten als typisches Syndrom des Smith-Magenis-Syndrom – guter therapeutischer Effekt von Aripiprazol.** *Medizin für Menschen mit geistiger oder mehrfacher Behinderung*; Edition Bentheim 2010(2), 45-51.

Li F, Shen Y, Koehler U, et al. **Interstitial microduplication of Xp22.31: Causative of intellectual disability or benign copy number variant?** *Eur J Med Genet.* 2010 Feb 2.

Bulst S, Abicht A, Holinski-Feder E, et al. **In vitro supplementation with dAMP/dGMP leads to partial restoration of mtDNA levels in mitochondrial depletion syndromes.** *Hum Mol Genet.* 2009;18(9):1590-1599. doi:[10.1093/hmg/ddp074](https://doi.org/10.1093/hmg/ddp074)

Mayer V, Schoen U, Holinski-Feder E, Koehler U, Thalhammer S. **Single cell analysis of mutations in the APC gene.** *Fetal Diagn Ther.* 2009;26(3):148-156. doi:[10.1159/000248721](https://doi.org/10.1159/000248721)

Woide D, Mayer V, Wachtmeister T, Hoehn N, Zink A, Koehler U, Thalhammer S. **Single particle adsorbing transfer system.** *Biomed Microdevices.* 2009 11(3): 609-614.

Tangcharoen T, Jahnke C, Koehler U, et al. **Impact of heart rate variability in patients with normal sinus rhythm on image quality in coronary magnetic angiography.** *J Magn Reson Imaging.* 2008;28(1):74-79. doi:[10.1002/jmri.21426](https://doi.org/10.1002/jmri.21426)

Koehler U, Grabowski M, Bacher U, Holinski-Feder E. **A new interphase fluorescence in situ hybridization approach for genomic rearrangements involving MLH1 and MSH6 in hereditary nonpolyposis colorectal cancer-suspected mutation-negative patients.** *Cancer Genet Cytogenet.* 2007;175(1):81-84. doi:[10.1016/j.cancergencyto.2007.01.008](https://doi.org/10.1016/j.cancergencyto.2007.01.008)

S. Thalhammer S., Koehler U., et al. **Programmierbares, zytogenetisches Submikroliter-Chiplabor für molekular-diagnostische Anwendungen.** *GenomXPress* 2007 ISBN: 1617-562X, 1.07: 29-31.

Grabowski M, Mueller-Koch Y, Grasbon-Frodl E, et al. **Deletions account for 17% of pathogenic germline alterations in MLH1 and MSH2 in hereditary nonpolyposis colorectal cancer (HNPCC) families.** *Genet Test.* 2005;9(2):138-146. doi:[10.1089/gte.2005.9.138](https://doi.org/10.1089/gte.2005.9.138)

Stanyon R, Koehler U, Consigliere S. **Chromosome painting reveals that galagos have highly derived karyotypes.** *Am J Phys Anthropol.* 2002;117(4):319-326. doi:[10.1002/ajpa.10047](https://doi.org/10.1002/ajpa.10047)

Thalhammer S, Koehler U, Stark RW, Heckl WM. **GTG banding pattern on human metaphase chromosomes revealed by high resolution atomic-force microscopy.** *J Microsc.* 2001;202(Pt 3):464-467.

Consigliere S, Stanyon R, Koehler U, Arnold N, Wienberg J. **In situ hybridization (FISH) maps chromosomal homologies between *Alouatta belzebul* (Platyrrhini, Cebidae) and other primates and reveals extensive interchromosomal rearrangements between howler monkey genomes.** *Am J Primatol.* 1998;46(2):119-133. doi:[10.1002/\(SICI\)1098-2345\(1998\)46:2<119::AID-AJP2>3.0.CO;2-Z](https://doi.org/10.1002/(SICI)1098-2345(1998)46:2<119::AID-AJP2>3.0.CO;2-Z)

Bigoni F, Koehler U, Stanyon R, Ishida T, Wienberg J. **Fluorescence in situ hybridization establishes homology between human and silvered leaf monkey chromosomes, reveals reciprocal translocations between chromosomes homologous to human Y/5, 1/9, and 6/16, and delineates an X1X2Y1Y2/X1X1X2X2 sex-chromosome system.** *Am J Phys Anthropol.* 1997;102(3):315-327.
doi:[10.1002/\(SICI\)1096-8644\(199703\)102:3<315::AID-AJPA2>3.0.CO;2-U](https://doi.org/10.1002/(SICI)1096-8644(199703)102:3<315::AID-AJPA2>3.0.CO;2-U)

Bigoni F, Stanyon R, Koehler U, Morescalchi AM, Wienberg J. **Mapping homology between human and black and white colobine monkey chromosomes by fluorescent in situ hybridization.** *Am J Primatol.* 1997;42(4):289-298. doi:[10.1002/\(SICI\)1098-2345\(1997\)42:4<289::AID-AJP4>3.0.CO;2-T](https://doi.org/10.1002/(SICI)1098-2345(1997)42:4<289::AID-AJP4>3.0.CO;2-T)

Clark LN, Koehler U, Ward DC, Wienberg J, Hewitt JE. **Analysis of the organisation and localisation of the FSHD-associated tandem array in primates: implications for the origin and evolution of the 3.3 kb repeat family.** *Chromosoma.* 1996;105(3):180-189.

Consigliere S, Stanyon R, Koehler U, Agoramoorthy G, Wienberg J. **Chromosome painting defines genomic rearrangements between red howler monkey subspecies.** *Chromosome Res.* 1996;4(4):264-270.

Muller S, Koehler U, Wienberg J, et al. **Comparative fluorescence in situ hybridization mapping of primate chromosomes with Alu polymerase chain reaction generated probes from human/rodent somatic cell hybrids.** *Chromosome Res.* 1996;4(1):38-42.

Koehler U, Bigoni F, Wienberg J, Stanyon R. **Genomic reorganization in the concolor gibbon (*Hylobates concolor*) revealed by chromosome painting.** *Genomics*. 1995;30(2):287-292. doi:[10.1006/geno.1995.9875](https://doi.org/10.1006/geno.1995.9875)

Koehler U, Arnold N, Wienberg J, Tofanelli S, Stanyon R. **Genomic reorganization and disrupted chromosomal synteny in the siamang (*Hylobates syndactylus*) revealed by fluorescence in situ hybridization.** *Am J Phys Anthropol*. 1995;97(1):37-47. doi:[10.1002/ajpa.1330970104](https://doi.org/10.1002/ajpa.1330970104)

Stanyon R, Arnold N, Koehler U, Bigoni F, Wienberg J. **Chromosomal painting shows that “marked chromosomes” in lesser apes and Old World monkeys are not homologous and evolved by convergence.** *Cytogenet Cell Genet*. 1995;68(1-2):74-78. doi:[10.1159/000133894](https://doi.org/10.1159/000133894)

Koehler U, Abken H, et al. **A novel type of unstable homogeneously staining region with a head-to-tail arrangement: spontaneous decay and reintegration of DNA elements into a plethora of new chromosomal sites.** *Cytogenet Cell Genet*. 1995;68(1-2):33-38. doi:[10.1159/000133883](https://doi.org/10.1159/000133883)

Zastrow G, Koehler U, Muller F, et al. **Distinct mouse DNA sequences enable establishment and persistence of plasmid DNA polymers in mouse cells.** *Nucleic Acids Res*. 1989;17(5):1867-1879. doi:[10.1093/nar/17.5.1867](https://doi.org/10.1093/nar/17.5.1867)