

# Literaturverzeichnis

- [ABRAHAMS et al. 2002] ABRAHAMS, B.S., G. MAK, M. BERRY, D. PALMQUIST, J. SAIONZ, A. TAY, Y. TAN, S. BRENNER, E. SIMPSON und B. VENKATESH (2002). *Novel vertebrate genes and putative regulatory elements identified at kidney disease and NR2E1 loci.* Genomics, 80(1):45–53.
- [ACCARDI und MILLER 2004] ACCARDI, ALESSIO und C. MILLER (2004). *Secondary active transport mediated by a prokaryotic homologue of ClC Cl- channels.* Nature, 427(6977):803–807.
- [ALEXANDER und ELDER 1989] ALEXANDER, S. und J. ELDER (1989). *Endoglycosidases from Flavobacterium meningosepticum application to biological problems.* Methods Enzymol, 179:505–18.
- [ANCANS et al. 2001] ANCANS, J., D. J. TOBIN, M. J. HOOGDUIJN, N. P. SMIT, K. WAKAMATSU und A. J. THODY (2001). *Melanosomal pH controls rate of melanogenesis, eumelanin/phaeomelanin ratio and melanosome maturation in melanocytes and melanoma cells.* Exp Cell Res, 268(1):26–35.
- [ANGELI et al. 2006] ANGELI, A. DE, D. MONACHELLO, G. EPHRITIKHINE, J. M. FRACHISSE, S. THOMINE, F. GAMBALE und H. BARBIER-BRYGOO (2006). *The nitrate/proton antiporter AtCLCa mediates nitrate accumulation in plant vacuoles.* Nature, 442(7105):939–942.
- [BAHR und BENDISKE 2002] BAHR, BEN A und J. BENDISKE (2002). *The neuropathogenic contributions of lysosomal dysfunction.* J Neurochem, 83(3):481–489.
- [BARON et al. 1985] BARON, R., L. NEFF, D. LOUVARD und P. COURTOY (1985). *Cell-mediated extracellular acidification and bone resorption: evidence for a low pH in resorbing lacunae and localization of a 100-kD lysosomal membrane protein at the osteoclast ruffled border.* J Cell Biol, 101(6):2210–22.
- [BARTHELMES et al. 2007] BARTHELMES, JENS, C. EBELING, A. CHANG, I. SCHOMBURG und D. SCHOMBURG (2007). *BRENDA, AMENDA and FRENDA: the enzyme information system in 2007.* Nucleic Acids Res, 35(Database issue):D511–D514.
- [BATEMAN 1997] BATEMAN, A. (1997). *The structure of a domain common to archaeobacteria and the homocystinuria disease protein.* Trends Biochem Sci, 22(1):12–13.

- [BAUDHUIN et al. 1965] BAUDHUIN, P., H. BEAUFAY und C. D. DUVE (1965). *Combined biochemical and morphological study of particulate fractions from rat liver: Analysis of preparations enriched in lysosomes or in particles containing urate oxidase, D-amino acid oxidase, and catalase..* J Cell Biol, 26(1):219–243.
- [BENNETT und LAMOREUX 2003] BENNETT, DOROTHY C und M. L. LAMOREUX (2003). *The color loci of mice—a genetic century..* Pigment Cell Res, 16(4):333–344.
- [BENNETTS et al. 2005] BENNETTS, BRETT, G. Y. RYCHKOV, H.-L. NG, C. J. MORTON, D. STAPLETON, M. W. PARKER und B. A. CROMER (2005). *Cytoplasmic ATP-sensing domains regulate gating of skeletal muscle ClC-1 chloride channels..* J Biol Chem, 280(37):32452–32458.
- [BÉGUIN et al. 2000] BÉGUIN, P., U. HASLER, O. STAUB und K. GEERING (2000). *Endoplasmic reticulum quality control of oligomeric membrane proteins: topogenic determinants involved in the degradation of the unassembled Na,K-ATPase alpha subunit and in its stabilization by beta subunit assembly..* Mol Biol Cell, 11(5):1657–1672.
- [BHATNAGAR et al. 1993] BHATNAGAR, V., S. ANJIAH, N. PURI, B. N. DARSHANAM und A. RAMAIAH (1993). *pH of melanosomes of B 16 murine melanoma is acidic: its physiological importance in the regulation of melanin biosynthesis..* Arch Biochem Biophys, 307(1):183–192.
- [BICHET et al. 2000] BICHET, D., V. CORNET, S. GEIB, E. CARLIER, S. VOLSEN, T. HOSHI, Y. MORI und M. D. WAARD (2000). *The I-II loop of the Ca<sup>2+</sup> channel alpha1 subunit contains an endoplasmic reticulum retention signal antagonized by the beta subunit..* Neuron, 25(1):177–190.
- [BIRKENHÄGER et al. 2001] BIRKENHÄGER, R., E. OTTO, M. J. SCHÜRSMANN, M. VOLLMER, E. M. RUF, I. MAIER-LUTZ, F. BEEKMANN, A. FEKETE, H. OM-RAN, D. FELDMANN, D. V. MILFORD, N. JECK, M. KONRAD, D. LANDAU, N. V. KNOERS, C. ANTIGNAC, R. SUDBRAK, A. KISPERS und F. HILDEBRANDT (2001). *Mutation of BSND causes Bartter syndrome with sensorineural deafness and kidney failure..* Nat Genet, 29(3):310–314.
- [BIRNBOIM und DOLY 1979] BIRNBOIM, H.C. und J. DOLY (1979). *A rapid alkaline extraction procedure for screening recombinant plasmid DNA..* Nucleic Acids Res, 7(6):1513–23.
- [BORDALLO et al. 1998] BORDALLO, J., R. PLEMPER, A. FINGER und D. WOLF (1998). *Der3p/Hrd1p is required for endoplasmic reticulum-associated degradation of misfolded luminal and integral membrane proteins..* Mol Biol Cell, 9(1):209–22.
- [BOSSCHE et al. 2006] BOSSCHE, KAROLIEN VAN DEN, J.-M. NAEYAERT und J. LAMBERT (2006). *The quest for the mechanism of melanin transfer..* Traffic, 7(7):769–778.

- [BRANDT und JENTSCH 1995] BRANDT, S. und T. J. JENTSCH (1995). *CLC-6 and CLC-7 are two novel broadly expressed members of the CLC chloride channel family.* FEBS Lett, 377(1):15–20.
- [BRICE et al. 1997] BRICE, N. L., N. S. BERROW, V. CAMPBELL, K. M. PAGE, K. BRICKLEY, I. TEDDER und A. C. DOLPHIN (1997). *Importance of the different beta subunits in the membrane expression of the alpha1A and alpha2 calcium channel subunits: studies using a depolarization-sensitive alpha1A antibody.* Eur J Neurosci, 9(4):749–759.
- [BURCHILL et al. 1989] BURCHILL, S. A., R. VIRDEN und A. J. THODY (1989). *Regulation of tyrosinase synthesis and its processing in the hair follicular melanocytes of the mouse during eumelanogenesis and phaeomelanogenesis.* J Invest Dermatol, 93(2):236–240.
- [CAI et al. 2006] CAI, SHI-QING, K. H. PARK und F. SESTI (2006). *An evolutionarily conserved family of accessory subunits of K<sup>+</sup> channels.* Cell Biochem Biophys, 46(1):91–99.
- [CAMPOS-XAVIER et al. 2003] CAMPOS-XAVIER, A.B., J. SARAIVA, L. RIBEIRO, A. MUNNICH und V. CORMIER-DAIRE (2003). *Chloride channel 7 (CLCN7) gene mutations in intermediate autosomal recessive osteopetrosis.* Hum Genet, 112(2):186–9.
- [CARR et al. 2003] CARR, G., N. SIMMONS und J. SAYER (2003). *A role for CBS domain 2 in trafficking of chloride channel CLC-5.* Biochem Biophys Res Commun, 310(2):600–5.
- [CHALHOUB et al. 2003] CHALHOUB, N., N. BENACHENHOU, V. RAJAPUROHITAM, M. PATA, M. FERRON, A. FRATTINI, A. VILLA und J. VACHER (2003). *Grey-lethal mutation induces severe malignant autosomal recessive osteopetrosis in mouse and human.* Nat Med, 9(4):399–406.
- [CIGIC und PAIN 1999] CIGIC, B. und R. H. PAIN (1999). *Location of the binding site for chloride ion activation of cathepsin C.* Eur J Biochem, 264(3):944–951.
- [CLEIREN et al. 2001] CLEIREN, E., O. BÉNICHOU, E. V. HUL, J. GRAM, J. BOLLEERSLEV, F. R. SINGER, K. BEAVERSON, A. ALEDO, M. P. WHYTE, T. YONEYAMA, M. C. DEVERNEJOU und W. V. HUL (2001). *Albers-Schönberg disease (autosomal dominant osteopetrosis, type II) results from mutations in the CLCN7 chloride channel gene.* Hum Mol Genet, 10(25):2861–2867.
- [COHEN 2006] COHEN, M. MICHAEL (2006). *The new bone biology: pathologic, molecular, and clinical correlates.* Am J Med Genet A, 140(23):2646–2706.

- [DEVI et al. 1987] DEVI, C. C., R. K. TRIPATHI und A. RAMAIAH (1987). *pH-dependent interconvertible allosteric forms of murine melanoma tyrosinase. Physiological implications..* Eur J Biochem, 166(3):705–711.
- [DHANI et al. 2003] DHANI, S.U., R. MOHAMMAD-PANAH, N. AHMED, C. ACKERLEY, M. RAMJEESINGH und C. BEAR (2003). *Evidence for a functional interaction between the CIC-2 chloride channel and the retrograde motor dynein complex..* J Biol Chem, 278(18):16262–70.
- [DOLPHIN 2003] DOLPHIN, A.C. (2003). *Beta subunits of voltage-gated calcium channels..* J Bioenerg Biomembr, 35(6):599–620.
- [DUNNE und PETERSEN 1986] DUNNE, M. J. und O. H. PETERSEN (1986). *Intracellular ADP activates K<sup>+</sup> channels that are inhibited by ATP in an insulin-secreting cell line..* FEBS Lett, 208(1):59–62.
- [DUNPHY und ROTHMAN 1985] DUNPHY, W. G. und J. E. ROTHMAN (1985). *Compartmental organization of the Golgi stack..* Cell, 42(1):13–21.
- [DUTZLER 2007] DUTZLER, RAIMUND (2007). *A structural perspective on CIC channel and transporter function..* FEBS Lett.
- [DUTZLER et al. 2002] DUTZLER, RAIMUND, E. B. CAMPBELL, M. CADENE, B. T. CHAIT und R. MACKINNON (2002). *X-ray structure of a CIC chloride channel at 3.0 Å reveals the molecular basis of anion selectivity..* Nature, 415(6869):287–294.
- [ELDER und ALEXANDER 1982] ELDER, J.H. und S. ALEXANDER (1982). *endo-beta-N-acetylglucosaminidase F: endoglycosidase from Flavobacterium meningosepticum that cleaves both high-mannose and complex glycoproteins..* Proc Natl Acad Sci USA, 79(15):4540–4.
- [ELLGAARD und HELENIUS 2003] ELLGAARD, LARS und A. HELENIUS (2003). *Quality control in the endoplasmic reticulum..* Nat Rev Mol Cell Biol, 4(3):181–191.
- [ESKELINEN et al. 2003] ESKELINEN, EEVA-LIISA, Y. TANAKA und P. SAFTIG (2003). *At the acidic edge: emerging functions for lysosomal membrane proteins..* Trends Cell Biol, 13(3):137–145.
- [ESTEVEZ et al. 2004] ESTEVEZ, R., M. PUSCH, C. FERRER-COSTA, M. OROZCO und T. JENTSCH (2004). *Functional and structural conservation of CBS domains from CLC channels..* J Physiol.
- [ESTÉVEZ et al. 2001] ESTÉVEZ, R., T. BOETTGER, V. STEIN, R. BIRKENHÄGER, E. OTTO, F. HILDEBRANDT und T. J. JENTSCH (2001). *Barttin is a Cl<sup>-</sup> channel beta-subunit crucial for renal Cl<sup>-</sup> reabsorption and inner ear K<sup>+</sup> secretion..* Nature, 414(6863):558–561.

- [FAUNDEZ und HARTZELL 2004] FAUNDEZ, VICTOR und H. C. HARTZELL (2004). *Intracellular chloride channels: determinants of function in the endosomal pathway.* Sci STKE, 2004(233):re8.
- [FISCHER et al. 2003] FISCHER, THIERRY, L. D. VRIES, T. MEERLOO und M. G. FARQUHAR (2003). *Promotion of G alpha i3 subunit down-regulation by GIPN, a putative E3 ubiquitin ligase that interacts with RGS-GAIP.* Proc Natl Acad Sci U S A, 100(14):8270–8275.
- [FLORES et al. 2006] FLORES, CARLOS A, M. I. NIEMEYER, F. V. SEPÚLVEDA und L. P. CID (2006). *Two splice variants derived from a Drosophila melanogaster candidate ClC gene generate ClC-2-type Cl- channels.* Mol Membr Biol, 23(2):149–156.
- [FRATTINI et al. 2000] FRATTINI, A., P. ORCHARD, C. SOBACCHI, S. GILIANI, M. ABINUN, J. MATTSSON, D. KEELING, A. ANDERSSON, P. WALLBRANDT, L. ZECCA, L. NOTARANGELO, P. VEZZONI und A. VILLA (2000). *Defects in TCIRG1 subunit of the vacuolar proton pump are responsible for a subset of human autosomal recessive osteopetrosis.* Nat Genet, 25(3):343–6.
- [FRATTINI et al. 2003] FRATTINI, A., A. PANGRAZIO, L. SUSANI, C. SOBACCHI, M. MIROLO, M. ABINUN, M. ANDOLINA, A. FLANAGAN, E. HORWITZ, E. MIHCI, L. NOTARANGELO, U. RAMENGI, A. TETI, J. VAN HOVE, D. VUJIC, T. YOUNG, A. ALBERTINI, P. ORCHARD, P. VEZZONI und A. VILLA (2003). *Chloride channel CLCN7 mutations are responsible for severe recessive, dominant, and intermediate osteopetrosis.* J Bone Miner Res, 18(10):1740–7.
- [FUJITA et al. 1994] FUJITA, N., H. MORI, T. YURA und A. ISHIHAMA (1994). *Systematic sequencing of the Escherichia coli genome: analysis of the 2.4-4.1 min (110,917-193,643 bp) region.* Nucleic Acids Res, 22(9):1637–1639.
- [FUKUDA 1991] FUKUDA, M. (1991). *Lysosomal membrane glycoproteins. Structure, biosynthesis, and intracellular trafficking.* J Biol Chem, 266(32):21327–21330.
- [FURUKAWA et al. 2002] FURUKAWA, TETSUSHI, T. OGURA, Y.-J. ZHENG, H. TSUCHIYA, H. NAKAYA, Y. KATAYAMA und N. INAGAKI (2002). *Phosphorylation and functional regulation of ClC-2 chloride channels expressed in Xenopus oocytes by M cyclin-dependent protein kinase.* J Physiol, 540(Pt 3):883–893.
- [GALBRAITH 1964] GALBRAITH, D. B. (1964). *THE AGOUTI PIGMENT PATTERN OF THE MOUSE: A QUANTITATIVE AND EXPERIMENTAL STUDY.* J Exp Zool, 155:71–90.
- [GENTZSCH et al. 2003] GENTZSCH, M., L. CUI, A. MENGOS, X. CHANG, J. CHEN und J. RIORDAN (2003). *The PDZ-binding chloride channel ClC-3B localizes to the Golgi and associates with cystic fibrosis transmembrane conductance regulator-interacting PDZ proteins.* J Biol Chem, 278(8):6440–9.

- [GERRITSEN et al. 1994a] GERRITSEN, E. J., J. M. VOSSEN, A. FASTH, W. FRIEDRICH, G. MORGAN, A. PADMOS, A. VELLODI, O. PORRAS, A. O'MEARA und F. PORTA (1994a). *Bone marrow transplantation for autosomal recessive osteopetrosis. A report from the Working Party on Inborn Errors of the European Bone Marrow Transplantation Group.* J Pediatr, 125(6 Pt 1):896–902.
- [GERRITSEN et al. 1994b] GERRITSEN, E.J., J. VOSSEN, A. FASTH, W. FRIEDRICH, G. MORGAN, A. PADMOS, A. VELLODI, O. PORRAS, A. O'MEARA, F. PORTA und ET AL. (1994b). *Bone marrow transplantation for autosomal recessive osteopetrosis. A report from the Working Party on Inborn Errors of the European Bone Marrow Transplantation Group.* J Pediatr, 125(6 Pt 1):896–902.
- [GILCHRIST et al. 2006] GILCHRIST, ANNALYN, C. E. AU, J. HIDING, A. W. BELL, J. FERNANDEZ-RODRIGUEZ, S. LESIMPLE, H. NAGAYA, L. ROY, S. J. C. GOSLINE, M. HALLETT, J. PAIEMENT, R. E. KEARNEY, T. NILSSON und J. J. M. BERGERON (2006). *Quantitative proteomics analysis of the secretory pathway.* Cell, 127(6):1265–1281.
- [GÜNTHER et al. 2003] GÜNTHER, WILLY, N. PIWON und T. J. JENTSCH (2003). *The CIC-5 chloride channel knock-out mouse - an animal model for Dent's disease.* Pflügers Arch, 445(4):456–462.
- [GOEBEL und WISNIEWSKI 2004] GOEBEL, HANS H und K. E. WISNIEWSKI (2004). *Current state of clinical and morphological features in human NCL.* Brain Pathol, 14(1):61–69.
- [GOLDENTHAL et al. 1985] GOLDENTHAL, K.L., K. HEDMAN, J. CHEN, J. AUGUST und M. WILLINGHAM (1985). *Postfixation detergent treatment for immunofluorescence suppresses localization of some integral membrane proteins.* J Histochem Cytochem, 33(8):813–20.
- [GREENE et al. 1993] GREENE, J. R., N. H. BROWN, B. J. DIDOMENICO, J. KAPLAN und D. J. EIDE (1993). *The GEF1 gene of Saccharomyces cerevisiae encodes an integral membrane protein; mutations in which have effects on respiration and iron-limited growth.* Mol Gen Genet, 241(5-6):542–553.
- [GRUENBERG und STENMARK 2004] GRUENBERG, JEAN und H. STENMARK (2004). *The biogenesis of multivesicular endosomes.* Nat Rev Mol Cell Biol, 5(4):317–323.
- [GRUENBERG 1935] GRUENBERG, H. (1935). *A New Sub-Lethal Colour Mutation in the House Mouse.* Royal Society of London Proceedings Series B, 118:321–342.
- [GRUENBERG 1936] GRUENBERG, H (1936). *Grey-lethal, a new mutation in the house mouse.* J. Heredity, 27:105–109.

- [HALTIA 2003] HALTIA, MATTI (2003). *The neuronal ceroid-lipofuscinoses..* J Neuro-pathol Exp Neurol, 62(1):1–13.
- [HALTIA 2006] HALTIA, MATTI (2006). *The neuronal ceroid-lipofuscinoses: from past to present..* Biochim Biophys Acta, 1762(10):850–856.
- [HAN et al. 2007] HAN, H.-Y., J.-R. LEE, W.-A. XU, M.-J. HAHN, J.-M. YANG und Y.-D. PARK (2007). *Effect of cl(-) on tyrosinase: complex inhibition kinetics and biochemical implication..* J Biomol Struct Dyn, 25(2):165–172.
- [HARA-CHIKUMA et al. 2005] HARA-CHIKUMA, MARIKO, B. YANG, N. D. SONAWANE, S. SASAKI, S. UCHIDA und A. S. VERKMAN (2005). *CLC-3 chloride channels facilitate endosomal acidification and chloride accumulation..* J Biol Chem, 280(2):1241–1247.
- [HARDING und GEUZE 1993] HARDING, C.V. und H. GEUZE (1993). *Immunogenic peptides bind to class II MHC molecules in an early lysosomal compartment..* J Immunol, 151(8):3988–98.
- [HAUSMANN et al. 2007] HAUSMANN, GEORGE, C. BÄNZIGER und K. BASLER (2007). *Helping Wingless take flight: how WNT proteins are secreted..* Nat Rev Mol Cell Biol, 8(4):331–336.
- [HEARING und EKEL 1976] HEARING, V. J. und T. M. EKEL (1976). *Mammalian tyrosinase. A comparison of tyrosine hydroxylation and melanin formation..* Biochem J, 157(3):549–557.
- [HEBEISEN et al. 2004] HEBEISEN, S., A. BIELA, B. GIESE, G. MULLER-NEWEN, P. HIDALGO und C. FAHLKE (2004). *The role of the carboxy-terminus in CLC chloride channel function..* J Biol Chem. aheadofprint.
- [HECHENBERGER et al. 1996] HECHENBERGER, M., B. SCHWAPPACH, W. N. FISCHER, W. B. FROMMER, T. J. JENTSCH und K. STEINMEYER (1996). *A family of putative chloride channels from Arabidopsis and functional complementation of a yeast strain with a CLC gene disruption..* J Biol Chem, 271(52):33632–33638.
- [HIRSCHBERG und SNIDER 1987] HIRSCHBERG, C. B. und M. D. SNIDER (1987). *Topography of glycosylation in the rough endoplasmic reticulum and Golgi apparatus..* Annu Rev Biochem, 56:63–87.
- [HRYCIW et al. 2006] HRYCIW, DEANNE H, J. EKBERG, C. FERGUSON, A. LEE, D. WANG, R. G. PARTON, C. A. POLLOCK, C. C. YUN und P. PORONNIK (2006). *Regulation of albumin endocytosis by PSD95/Dlg/ZO-1 (PDZ) scaffolds. Interaction of Na<sup>+</sup>-H<sup>+</sup> exchange regulatory factor-2 with CLC-5..* J Biol Chem, 281(23):16068–16077.

- [HRYCIW et al. 2004] HRYCIW, DEANNE H, J. EKBERG, A. LEE, I. L. LENSINK, S. KUMAR, W. B. GUGGINO, D. I. COOK, C. A. POLLOCK und P. PORONNIK (2004). *Nedd4-2 functionally interacts with CLC-5: involvement in constitutive albumin endocytosis in proximal tubule cells.* J Biol Chem, 279(53):54996–55007.
- [HRYCIW et al. 2003] HRYCIW, D.H., Y. WANG, O. DEVUYST, C. POLLOCK, P. PORONNIK und W. GUGGINO (2003). *Cofilin interacts with CLC-5 and regulates albumin uptake in proximal tubule cell lines.* J Biol Chem, 278(41):40169–76.
- [HUNT et al. 1995] HUNT, G., S. KYNE, K. WAKAMATSU, S. ITO und A. J. THODY (1995). *Nle4DPhe7 alpha-melanocyte-stimulating hormone increases the eumelanin:phaeomelanin ratio in cultured human melanocytes.* J Invest Dermatol, 104(1):83–85.
- [JENTSCH et al. 1990] JENTSCH, T. J., K. STEINMEYER und G. SCHWARZ (1990). *Primary structure of Torpedo marmorata chloride channel isolated by expression cloning in Xenopus oocytes.* Nature, 348(6301):510–514.
- [JENTSCH 2007] JENTSCH, THOMAS J (2007). *Chloride and the endosomal-lysosomal pathway: emerging roles of CLC chloride transporters.* J Physiol, 578(Pt 3):633–640.
- [JENTSCH et al. 2005] JENTSCH, THOMAS J, M. POËT, J. C. FUHRMANN und A. A. ZDEBIK (2005). *Physiological functions of CLC Cl<sup>-</sup> channels gleaned from human genetic disease and mouse models.* Annu Rev Physiol, 67:779–807.
- [JENTSCH et al. 2002] JENTSCH, THOMAS J, V. STEIN, F. WEINREICH und A. A. ZDEBIK (2002). *Molecular structure and physiological function of chloride channels.* Physiol Rev, 82(2):503–568.
- [KASPER et al. 2005] KASPER, DAGMAR, R. PLANELLS-CASES, J. C. FUHRMANN, O. SCHEEL, O. ZEITZ, K. RUETHER, A. SCHMITT, M. POËT, R. STEINFELD, M. SCHWEIZER, U. KORNAK und T. J. JENTSCH (2005). *Loss of the chloride channel CLC-7 leads to lysosomal storage disease and neurodegeneration.* EMBO J, 24(5):1079–1091.
- [KOCH et al. 1992] KOCH, M. C., K. STEINMEYER, C. LORENZ, K. RICKER, F. WOLF, M. OTTO, B. ZOLL, F. LEHMANN-HORN, K. H. GRZESCHIK und T. J. JENTSCH (1992). *The skeletal muscle chloride channel in dominant and recessive human myotonia.* Science, 257(5071):797–800.
- [KORNAK et al. 2001] KORNAK, U., D. KASPER, M. R. BÖSL, E. KAISER, M. SCHWEIZER, A. SCHULZ, W. FRIEDRICH, G. DELLING und T. J. JENTSCH (2001). *Loss of the CLC-7 chloride channel leads to osteopetrosis in mice and man.* Cell, 104(2):205–215.

- [KORNAK et al. 2000] KORNAK, U., A. SCHULZ, W. FRIEDRICH, S. UHLHAAS, B. KREMENS, T. VOIT, C. HASAN, U. BODE, T. J. JENTSCH und C. KUBISCH (2000). *Mutations in the  $\alpha 3$  subunit of the vacuolar H(+)-ATPase cause infantile malignant osteopetrosis..* Hum Mol Genet, 9(13):2059–2063.
- [KORNFELD und KORNFELD 1985] KORNFELD, R. und S. KORNFELD (1985). *Assembly of asparagine-linked oligosaccharides..* Annu Rev Biochem, 54:631–664.
- [KORNFELD und MELLMAN 1989] KORNFELD, S. und I. MELLMAN (1989). *The biogenesis of lysosomes..* Annu Rev Cell Biol, 5:483–525.
- [KUNDRA und KORNFELD 1999] KUNDRA, R. und S. KORNFELD (1999). *Asparagine-linked oligosaccharides protect Lamp-1 and Lamp-2 from intracellular proteolysis..* J Biol Chem, 274(43):31039–31046.
- [KYTE und DOOLITTLE 1982] KYTE, J. und R. F. DOOLITTLE (1982). *A simple method for displaying the hydropathic character of a protein..* J Mol Biol, 157(1):105–132.
- [LAEMMLI 1970] LAEMMLI, U.K. (1970). *Cleavage of structural proteins during the assembly of the head of bacteriophage T4..* Nature, 227(259):680–5.
- [LETIZIA et al. 2004] LETIZIA, C., A. TARANTA, S. MIGLIACCIO, C. CALIUMI, D. DIACINTI, E. DELFINI, E. D'ERASMO, M. IACOBINI, M. ROGGINI, O. ALBAGHA, S. RALSTON und A. TETI (2004). *Type II benign osteopetrosis (Albers-Schonberg disease) caused by a novel mutation in CLCN7 presenting with unusual clinical manifestations..* Calcif Tissue Int, 74(1):42–6.
- [LEWIS et al. 1985] LEWIS, V., S. A. GREEN, M. MARSH, P. VIHKO, A. HELENIUS und I. MELLMAN (1985). *Glycoproteins of the lysosomal membrane..* J Cell Biol, 100(6):1839–1847.
- [LI et al. 1999] LI, Y.P., W. CHEN, Y. LIANG, E. LI und P. STASHENKO (1999). *Atp6i-deficient mice exhibit severe osteopetrosis due to loss of osteoclast-mediated extracellular acidification..* Nat Genet, 23(4):447–51.
- [LI et al. 1996] LI, Y.P., W. CHEN und P. STASHENKO (1996). *Molecular cloning and characterization of a putative novel human osteoclast-specific 116-kDa vacuolar proton pump subunit..* Biochem Biophys Res Commun, 218(3):813–21.
- [LLOYD et al. 1997] LLOYD, S. E., W. GUNTHER, S. H. PEARCE, A. THOMSON, M. L. BIANCHI, M. BOSIO, I. W. CRAIG, S. E. FISHER, S. J. SCHEINMAN, O. WRONG, T. J. JENTSCH und R. V. THAKKER (1997). *Characterisation of renal chloride channel, CLCN5, mutations in hypercalciuric nephrolithiasis (kidney stones) disorders..* Hum Mol Genet, 6(8):1233–1239.

- [LORENZ et al. 1996] LORENZ, C., M. PUSCH und T. J. JENTSCH (1996). *Heteromultimeric CLC chloride channels with novel properties..* Proc Natl Acad Sci U S A, 93(23):13362–13366.
- [LUDEWIG et al. 1996] LUDEWIG, U., M. PUSCH und T. J. JENTSCH (1996). *Two physically distinct pores in the dimeric CLC-0 chloride channel..* Nature, 383(6598):340–343.
- [LURIN et al. 2000] LURIN, C., J. GÜCLÜ, C. CHENICLET, J. P. CARDE, H. BARBIER-BRYGOO und C. MAUREL (2000). *CLC-Nt1, a putative chloride channel protein of tobacco, co-localizes with mitochondrial membrane markers..* Biochem J, 348 Pt 2:291–295.
- [LUZIO et al. 2007] LUZIO, J. PAUL, P. R. PRYOR und N. A. BRIGHT (2007). *Lysosomes: fusion and function..* Nat Rev Mol Cell Biol, 8(8):622–632.
- [MARANDA et al. 2007] MARANDA, B., G. CHABOT, J.-C. DÉCARIE, M. PATA, B. AZEDDINE, A. MOREAU und J. VACHER (2007). *Clinical and Cellular Manifestations of OSTM1 Related Infantile Osteopetrosis..* J Bone Miner Res.
- [MARKOVIC und DUTZLER 2007] MARKOVIC, SANDRA und R. DUTZLER (2007). *The Structure of the Cytoplasmic Domain of the Chloride Channel CLC-Ka Reveals a Conserved Interaction Interface..* Structure, 15(6):715–725.
- [MARLES et al. 2003] MARLES, LEE K, E. M. PETERS, D. J. TOBIN, N. A. HIBBERTS und K. U. SCHALLREUTER (2003). *Tyrosine hydroxylase isoenzyme I is present in human melanosomes: a possible novel function in pigmentation..* Exp Dermatol, 12(1):61–70.
- [MELLMAN et al. 1986] MELLMAN, I., R. FUCHS und A. HELENIUS (1986). *Acidification of the endocytic and exocytic pathways..* Annu Rev Biochem, 55:663–700.
- [MEYER et al. 2007] MEYER, SEBASTIAN, S. SAVARESI, I. C. FORSTER und R. DUTZLER (2007). *Nucleotide recognition by the cytoplasmic domain of the human chloride transporter CLC-5..* Nat Struct Mol Biol, 14(1):60–67.
- [MICHELSEN et al. 2005] MICHELSEN, KAI, H. YUAN und B. SCHWAPPACH (2005). *Hide and run. Arginine-based endoplasmic-reticulum-sorting motifs in the assembly of heteromultimeric membrane proteins..* EMBO Rep, 6(8):717–722.
- [MICHIGAMI et al. 2002] MICHIGAMI, T., T. KAGEYAMA, K. SATOMURA, M. SHIMA, K. YAMAOKA, M. NAKAYAMA und K. OZONO (2002). *Novel mutations in the  $\alpha 3$  subunit of vacuolar H(+)-adenosine triphosphatase in a Japanese patient with infantile malignant osteopetrosis..* Bone, 30(2):436–9.

- [MILLER et al. 1993] MILLER, A. L., V. NORTON, R. ROBERTSON, M. JENKS, R. Y. YEH und D. WRIGHT (1993). *Light and heavy lysosomes: characterization of N-acetyl-beta-D-hexosaminidase isolated from normal and I-cell disease lymphoblasts..* Glycobiology, 3(4):313–318.
- [MILLER und WHITE 1984] MILLER, C. und M. WHITE (1984). *Dimeric structure of single chloride channels from Torpedo electroplax..* Proc Natl Acad Sci U S A, 81(9):2772–5.
- [MOHAMMAD-PANAH et al. 2003] MOHAMMAD-PANAH, R., R. HARRISON, S. DHANI, C. ACKERLEY, L. HUAN, Y. WANG und C. BEAR (2003). *The chloride channel ClC-4 contributes to endosomal acidification and trafficking..* J Biol Chem, 278(31):29267–77.
- [MOLE 2004] MOLE, SARA (2004). *Neuronal ceroid lipofuscinoses (NCL)..* Eur J Paediatr Neurol, 8(2):101–103.
- [MULARI et al. 2003] MULARI, M.T., H. ZHAO, P. LAKKAKORPI und H. VAANANEN (2003). *Osteoclast ruffled border has distinct subdomains for secretion and degraded matrix uptake..* Traffic, 4(2):113–25.
- [NAKAHIRA et al. 1996] NAKAHIRA, K., G. SHI, K. J. RHODES und J. S. TRIMMER (1996). *Selective interaction of voltage-gated K<sup>+</sup> channel beta-subunits with alpha-subunits..* J Biol Chem, 271(12):7084–7089.
- [NAKATSU und OHNO 2003] NAKATSU, FUBITO und H. OHNO (2003). *Adaptor protein complexes as the key regulators of protein sorting in the post-Golgi network..* Cell Struct Funct, 28(5):419–429.
- [NEISS 1984] NEISS, W. F. (1984). *A coat of glycoconjugates on the inner surface of the lysosomal membrane in the rat kidney..* Histochemistry, 80(6):603–608.
- [NICHOLS 2006] NICHOLS, COLIN G (2006). *KATP channels as molecular sensors of cellular metabolism..* Nature, 440(7083):470–476.
- [NISHI und FORGAC 2002] NISHI, T. und M. FORGAC (2002). *The vacuolar (H<sup>+</sup>)-ATPases–nature’s most versatile proton pumps..* Nat Rev Mol Cell Biol, 3(2):94–103.
- [OFFE 2005] OFFE, J. K. (2005). *Untersuchungen zu Lokalisationssignalen des Chloridkanals ClC-7 und Analyse eines ClC-3/ClC-7-Knockout-Mausmodells (Mus musculus).*
- [ORLOW 1995] ORLOW, S. J. (1995). *Melanosomes are specialized members of the lysosomal lineage of organelles..* J Invest Dermatol, 105(1):3–7.

- [PALOKANGAS et al. 1997] PALOKANGAS, H., M. MULARI und H. K. VÄÄNÄNEN (1997). *Endocytic pathway from the basal plasma membrane to the ruffled border membrane in bone-resorbing osteoclasts..* J Cell Sci, 110 ( Pt 15):1767–1780.
- [PANGRAZIO et al. 2006] PANGRAZIO, ALESSANDRA, P. L. POLIANI, A. MEGARBANE, G. LEFRANC, E. LANINO, M. D. ROCCO, F. RUCCI, F. LUCCHINI, M. RAVANINI, F. FACCHETTI, M. ABINUN, P. VEZZONI, A. VILLA und A. FRATTINI (2006). *Mutations in OSTM1 (grey lethal) define a particularly severe form of autosomal recessive osteopetrosis with neural involvement..* J Bone Miner Res, 21(7):1098–1105.
- [PARK et al. 2005] PARK, YONG-DOO, S.-Y. KIM, Y.-J. LYOU, J.-Y. LEE und J.-M. YANG (2005). *A new type of uncompetitive inhibition of tyrosinase induced by Cl-binding..* Biochimie, 87(11):931–937.
- [PICOLLO und PUSCH 2005] PICOLLO, ALESSANDRA und M. PUSCH (2005). *Chloride/proton antiporter activity of mammalian CLC proteins CLC-4 and CLC-5..* Nature, 436(7049):420–423.
- [PIWON et al. 2000] PIWON, N., W. GÜNTHER, M. SCHWAKE, M. R. BÖSL und T. J. JENTSCH (2000). *CLC-5 Cl<sup>-</sup>-channel disruption impairs endocytosis in a mouse model for Dent's disease..* Nature, 408(6810):369–373.
- [POËT et al. 2006] POËT, MALLORIE, U. KORNAK, M. SCHWEIZER, A. A. ZDEBIK, O. SCHEEL, S. HOELTER, W. WURST, A. SCHMITT, J. C. FUHRMANN, R. PLANELLS-CASES, S. E. MOLE, C. A. HÜBNER und T. J. JENTSCH (2006). *Lysosomal storage disease upon disruption of the neuronal chloride transport protein CLC-6..* Proc Natl Acad Sci U S A, 103(37):13854–13859.
- [PUNTERVOLL et al. 2003] PUNTERVOLL, P., R. LINDING, C. GEMUND, S. CHABANIS-DAVIDSON, M. MATTINGSDAL, S. CAMERON, D. MARTIN, G. AUSIELLO, B. BRANNETTI, A. COSTANTINI, F. FERRE, V. MASELLI, A. VIA, G. CESARENI, F. DIELLA, G. SUPERTI-FURGA, L. WYRWICZ, C. RAMU, C. MCGUIGAN, R. GUDAVALLI, I. LETUNIC, P. BORK, L. RYCHLEWSKI, B. KUSTER, M. HELMER-CITTERICH, W. HUNTER, R. AASLAND und T. GIBSON (2003). *ELM server: A new resource for investigating short functional sites in modular eukaryotic proteins..* Nucleic Acids Res, 31(13):3625–30.
- [PUSCH et al. 1995] PUSCH, M., U. LUDEWIG, A. REHFELDT und T. J. JENTSCH (1995). *Gating of the voltage-dependent chloride channel CLC-0 by the permeant anion..* Nature, 373(6514):527–531.
- [QORONFLEH et al. 2003] QORONFLEH, M., L. REN, D. EMERY, M. PERR und B. KAMBOOR (2003). *Use of Immunomatrix Methods to Improve Protein-Protein Interaction Detection..* J Biomed Biotechnol, 2003(5):291–298.

- [RAJAPUROHITAM et al. 2001] RAJAPUROHITAM, V., N. CHALHOUB, N. BENACHEN-HOU, L. NEFF, R. BARON und J. VACHER (2001). *The mouse osteopetrotic grey-lethal mutation induces a defect in osteoclast maturation/function..* Bone, 28(5):513–23.
- [RAMÍREZ et al. 2004] RAMÍREZ, ALFREDO, J. FAUPEL, I. GOEBEL, A. STILLER, S. BEYER, C. STÖCKLE, C. HASAN, U. BODE, U. KORNAK und C. KUBISCH (2004). *Identification of a novel mutation in the coding region of the grey-lethal gene OSTM1 in human malignant infantile osteopetrosis..* Hum Mutat, 23(5):471–476.
- [REEVES et al. 1979] REEVES, J.D., C. AUGUST, J. HUMBERT und W. WESTON (1979). *Host defense in infantile osteopetrosis..* Pediatrics, 64(2):202–6.
- [ROBERTS 2005] ROBERTS, REBECCA (2005). *Lysosomal cysteine proteases: structure, function and inhibition of cathepsins..* Drug News Perspect, 18(10):605–614.
- [ROBINSON und BONIFACINO 2001] ROBINSON, M. S. und J. S. BONIFACINO (2001). *Adaptor-related proteins..* Curr Opin Cell Biol, 13(4):444–453.
- [RUDDOCK und MOLINARI 2006] RUDDOCK, LLOYD W und M. MOLINARI (2006). *N-glycan processing in ER quality control..* J Cell Sci, 119(Pt 21):4373–4380.
- [RUTLEDGE et al. 2001] RUTLEDGE, E., L. BIANCHI, M. CHRISTENSEN, C. BOEHMER, R. MORRISON, A. BROSLAT, A. M. BELD, A. L. GEORGE, D. GREENSTEIN und K. STRANGE (2001). *CLH-3, a ClC-2 anion channel ortholog activated during meiotic maturation in C. elegans oocytes..* Curr Biol, 11(3):161–170.
- [SAEKI und OIKAWA 1985] SAEKI, H. und A. OIKAWA (1985). *Stimulation by ionophores of tyrosinase activity of mouse melanoma cells in culture..* J Invest Dermatol, 85(5):423–425.
- [SAITO et al. 2007] SAITO, MITSUYOSHI, P. I. HANSON und P. SCHLESINGER (2007). *Luminal chloride-dependent activation of endosome calcium channels: patch clamp study of enlarged endosomes..* J Biol Chem, 282(37):27327–27333.
- [SALO et al. 1997] SALO, J., P. LEHENKARI, M. MULARI, K. METSIKKO und H. VAANANEN (1997). *Removal of osteoclast bone resorption products by transcytosis..* Science, 276(5310):270–3.
- [SAMBROOK et al. 1989] SAMBROOK, J., E. FRITSCH und T. MANIATIS (1989). *Molecular cloning: A laboratory manual.* Cold Spring Harbour Laboratory Press, 2nd Aufl.
- [SANGER et al. 1977] SANGER, F., S. NICKLEN und A. COULSON (1977). *DNA sequencing with chain-terminating inhibitors..* Proc Natl Acad Sci U S A, 74(12):5463–7.

- [SCHEEL et al. 2005] SCHEEL, OLAF, A. A. ZDEBIK, S. LOURDEL und T. J. JENTSCH (2005). *Voltage-dependent electrogenic chloride/proton exchange by endosomal CLC proteins..* Nature, 436(7049):424–427.
- [SCHOLL et al. 2006] SCHOLL, UTE, S. HEBEISEN, A. G. H. JANSSEN, G. MÜLLER-NEWEN, A. ALEKOV und C. FAHLKE (2006). *Barttin modulates trafficking and function of ClC-K channels..* Proc Natl Acad Sci U S A, 103(30):11411–11416.
- [SCHOMBURG et al. 2000] SCHOMBURG, IDA, O. HOFMANN, C. BAENSCH, A. CHANG und D. SCHOMBURG (2000). *Enzyme data and metabolic information: BRENDA, a resource for research in biology, biochemistry, and medicine.* Gene Function & Disease, 1(3-4):109–118.
- [SCHRIEVER et al. 1999] SCHRIEVER, A. M., T. FRIEDRICH, M. PUSCH und T. J. JENTSCH (1999). *CLC chloride channels in Caenorhabditis elegans..* J Biol Chem, 274(48):34238–34244.
- [SCHROEDER et al. 2000] SCHROEDER, B. C., S. WALDEGGER, S. FEHR, M. BLEICH, R. WARTH, R. GREGER und T. J. JENTSCH (2000). *A constitutively open potassium channel formed by KCNQ1 and KCNE3..* Nature, 403(6766):196–199.
- [SCHWAKE et al. 2001] SCHWAKE, M., T. FRIEDRICH und T. J. JENTSCH (2001). *An internalization signal in ClC-5, an endosomal Cl-channel mutated in dent's disease..* J Biol Chem, 276(15):12049–12054.
- [SCHWAKE et al. 2003] SCHWAKE, MICHAEL, T. J. JENTSCH und T. FRIEDRICH (2003). *A carboxy-terminal domain determines the subunit specificity of KCNQ K+ channel assembly..* EMBO Rep, 4(1):76–81.
- [SCIMECA et al. 2000] SCIMECA, J.C., A. FRANCHI, C. TROJANI, H. PARRINELLO, J. GROSGEORGE, C. ROBERT, O. JAILLON, C. POIRIER, P. GAUDRAY und G. CARLE (2000). *The gene encoding the mouse homologue of the human osteoclast-specific 116-kDa V-ATPase subunit bears a deletion in osteosclerotic (oc/oc) mutants..* Bone, 26(3):207–13.
- [SCIMECA et al. 2003] SCIMECA, J.C., D. QUINCEY, H. PARRINELLO, D. ROMATET, J. GROSGEORGE, P. GAUDRAY, N. PHILIP, A. FISCHER und G. CARLE (2003). *Novel mutations in the TCIRG1 gene encoding the  $\alpha 3$  subunit of the vacuolar proton pump in patients affected by infantile malignant osteopetrosis..* Hum Mutat, 21(2):151–7.
- [SCOTT et al. 2004] SCOTT, J.W., S. HAWLEY, K. GREEN, M. ANIS, G. STEWART, G. SCULLION, D. NORMAN und D. HARDIE (2004). *CBS domains form energy-sensing modules whose binding of adenosine ligands is disrupted by disease mutations..* J Clin Invest, 113(2):274–84.

- [SHI et al. 1996] SHI, G., K. NAKAHIRA, S. HAMMOND, K. J. RHODES, L. E. SCHECHTER und J. S. TRIMMER (1996). *Beta subunits promote K<sup>+</sup> channel surface expression through effects early in biosynthesis.* Neuron, 16(4):843–852.
- [SIINTOLA et al. 2006] SIINTOLA, EIJA, A.-E. LEHESJOKI und S. E. MOLE (2006). *Molecular genetics of the NCLs – status and perspectives.* Biochim Biophys Acta, 1762(10):857–864.
- [SIMON et al. 1997] SIMON, D. B., R. S. BINDRA, T. A. MANSFIELD, C. NELSON-WILLIAMS, E. MENDONCA, R. STONE, S. SCHURMAN, A. NAYIR, H. ALPAY, A. BAKKALOGLU, J. RODRIGUEZ-SORIANO, J. M. MORALES, S. A. SANJAD, C. M. TAYLOR, D. PILZ, A. BREM, H. TRACHTMAN, W. GRISWOLD, G. A. RICHARD, E. JOHN und R. P. LIFTON (1997). *Mutations in the chloride channel gene, CLCNKB, cause Bartter's syndrome type III.* Nat Genet, 17(2):171–178.
- [SLOMINSKI et al. 2004] SLOMINSKI, ANDRZEJ, D. J. TOBIN, S. SHIBAHARA und J. WORTSMAN (2004). *Melanin pigmentation in mammalian skin and its hormonal regulation.* Physiol Rev, 84(4):1155–1228.
- [SLY et al. 1983] SLY, W.S., D. HEWETT-EMMETT, M. WHYTE, Y. YU und R. TASHIAN (1983). *Carbonic anhydrase II deficiency identified as the primary defect in the autosomal recessive syndrome of osteopetrosis with renal tubular acidosis and cerebral calcification.* Proc Natl Acad Sci U S A, 80(9):2752–6.
- [SONAWANE et al. 2002] SONAWANE, N. D., J. R. THIAGARAJAH und A. S. VERKMAN (2002). *Chloride concentration in endosomes measured using a ratioable fluorescent Cl<sup>-</sup> indicator: evidence for chloride accumulation during acidification.* J Biol Chem, 277(7):5506–5513.
- [SONAWANE und VERKMAN 2003] SONAWANE, N. D. und A. S. VERKMAN (2003). *Determinants of [Cl<sup>-</sup>] in recycling and late endosomes and Golgi complex measured using fluorescent ligands.* J Cell Biol, 160(7):1129–1138.
- [SOURATY et al. 2007] SOURATY, NOËLLE, P. NOUN, C. DJAMBAS-KHAYAT, E. CHOUERY, A. PANGRAZIO, A. VILLA, G. LEFRANC, A. FRATTINI und A. MÉGARBANÉ (2007). *Molecular study of six families originating from the Middle-East and presenting with autosomal recessive osteopetrosis.* Eur J Med Genet.
- [STEINMEYER et al. 1991] STEINMEYER, K., R. KLOCKE, C. ORTLAND, M. GRO-NEMEIER, H. JOCKUSCH, S. GRÜNDER und T. J. JENTSCH (1991). *Inactivation of muscle chloride channel by transposon insertion in myotonic mice.* Nature, 354(6351):304–308.
- [STENBECK 2002] STENBECK, GUDRUN (2002). *Formation and function of the ruffled border in osteoclasts.* Semin Cell Dev Biol, 13(4):285–292.

- [STOBRAWA et al. 2001] STOBRAWA, S. M., T. BREIDERHOFF, S. TAKAMORI, D. ENGEL, M. SCHWEIZER, A. A. ZDEBIK, M. R. BÖSL, K. RUETHER, H. JAHN, A. DRAGUHN, R. JAHN und T. J. JENTSCH (2001). *Disruption of ClC-3, a chloride channel expressed on synaptic vesicles, leads to a loss of the hippocampus..* Neuron, 29(1):185–196.
- [TAJIMA et al. 2007] TAJIMA, MASATO, A. HAYAMA, T. RAI, S. SASAKI und S. UCHIDA (2007). *Barttin binds to the outer lateral surface of the ClC-K2 chloride channel..* Biochem Biophys Res Commun, 362(4):858–864.
- [TAKAMORI et al. 2006] TAKAMORI, SHIGEO, M. HOLT, K. STENIUS, E. A. LEMKE, M. GRØNBORG, D. RIEDEL, H. URLAUB, S. SCHENCK, B. BRÜGGER, P. RINGLER, S. A. MÜLLER, B. RAMMNER, F. GRÄTER, J. S. HUB, B. L. D. GROOT, G. MIESKES, Y. MORIYAMA, J. KLINGAUF, H. GRUBMÜLLER, J. HEUSER, F. WIELAND und R. JAHN (2006). *Molecular anatomy of a trafficking organelle..* Cell, 127(4):831–846.
- [TEASDALE und JACKSON 1996] TEASDALE, R.D. und M. JACKSON (1996). *Signal-mediated sorting of membrane proteins between the endoplasmic reticulum and the golgi apparatus..* Annu Rev Cell Dev Biol, 12:27–54.
- [TEITELBAUM und ROSS 2003] TEITELBAUM, S.L. und F. ROSS (2003). *Genetic regulation of osteoclast development and function..* Nat Rev Genet, 4(8):638–49.
- [TOWNSEND et al. 1984] TOWNSEND, D., P. GUILLERY und R. A. KING (1984). *Optimized assay for mammalian tyrosinase (polyhydroxyl phenyloxidase)..* Anal Biochem, 139(2):345–352.
- [TOYOMURA et al. 2003] TOYOMURA, T., Y. MURATA, A. YAMAMOTO, T. OKA, G. SUN-WADA, Y. WADA und M. FUTAI (2003). *From lysosomes to the plasma membrane: localization of vacuolar-type H<sup>+</sup>-ATPase with the  $\alpha 3$  isoform during osteoclast differentiation..* J Biol Chem, 278(24):22023–30.
- [TOYOMURA et al. 2000] TOYOMURA, T., T. OKA, C. YAMAGUCHI, Y. WADA und M. FUTAI (2000). *Three subunit  $\alpha$  isoforms of mouse vacuolar H<sup>+</sup>-ATPase. Preferential expression of the  $\alpha 3$  isoform during osteoclast differentiation..* J Biol Chem, 275(12):8760–5.
- [VAANANEN et al. 2000] VAANANEN, H.K., H. ZHAO, M. MULARI und J. HALLEEN (2000). *The cell biology of osteoclast function..* J Cell Sci, 113 ( Pt 3):377–81.
- [VANOYE und GEORGE 2002] VANOYE, CARLOS G und A. L. GEORGE (2002). *Functional characterization of recombinant human ClC-4 chloride channels in cultured mammalian cells..* J Physiol, 539(Pt 2):373–383.

- [WAGUESPACK et al. 2007] WAGUESPACK, STEVEN G, S. L. HUI, L. A. DIMEGLIO und M. J. ECONS (2007). *Autosomal dominant osteopetrosis: clinical severity and natural history of 94 subjects with a chloride channel 7 gene mutation..* J Clin Endocrinol Metab, 92(3):771–778.
- [WAGUESPACK et al. 2003] WAGUESPACK, STEVEN G, D. L. KOLLER, K. E. WHITE, T. FISHBURN, G. CARN, K. A. BUCKWALTER, M. JOHNSON, M. KOCISKO, W. E. EVANS, T. FOROUD und M. J. ECONS (2003). *Chloride channel 7 (CLCN7) gene mutations and autosomal dominant osteopetrosis, type II..* J Bone Miner Res, 18(8):1513–1518.
- [WALDEGGER et al. 2002] WALDEGGER, SIEGFRIED, N. JECK, P. BARTH, M. PETERS, H. VITZTHUM, K. WOLF, A. KURTZ, M. KONRAD und H. W. SEYBERTH (2002). *Barttin increases surface expression and changes current properties of ClC-K channels..* Pflugers Arch, 444(3):411–418.
- [WALKER 1975] WALKER, D.G. (1975). *Bone resorption restored in osteopetrotic mice by transplants of normal bone marrow and spleen cells..* Science, 190:784–785.
- [WEINREICH und JENTSCH 2001] WEINREICH, F. und T. JENTSCH (2001). *Pores formed by single subunits in mixed dimers of different CLC chloride channels..* J Biol Chem, 276(4):2347–53.
- [WELLHAUSER et al. 2006] WELLHAUSER, LEIGH, H.-H. KUO, F. L. L. STRATFORD, M. RAMJEESINGH, L.-J. HUAN, W. LUONG, C. LI, C. M. DEBER und C. E. BEAR (2006). *Nucleotides bind to the C-terminus of ClC-5..* Biochem J, 398(2):289–294.
- [WILSON und VELLODI 2000] WILSON, C.J. und A. VELLODI (2000). *Autosomal recessive osteopetrosis: diagnosis, management, and outcome..* Arch Dis Child, 83(5):449–52.
- [ØYEHAUG et al. 2002] ØYEHAUG, LEIV, E. PLAHTÉ, D. I. VAGE und S. W. OMHOLT (2002). *The regulatory basis of melanogenic switching..* J Theor Biol, 215(4):449–468.
- [YOSHIKAWA et al. 2002] YOSHIKAWA, MOMONO, S. UCHIDA, J. EZAKI, T. RAI, A. HAYAMA, K. KOBAYASHI, Y. KIDA, M. NODA, M. KOIKE, Y. UCHIYAMA, F. MARUMO, E. KOMINAMI und S. SASAKI (2002). *CLC-3 deficiency leads to phenotypes similar to human neuronal ceroid lipofuscinosis..* Genes Cells, 7(6):597–605.
- [YU et al. 2003] YU, FRANK H, R. E. WESTENBROEK, I. SILOS-SANTIAGO, K. A. MCCORMICK, D. LAWSON, P. GE, H. FERRIERA, J. LILLY, P. S. DISTEFANO, W. A. CATTERALL, T. SCHEUER und R. CURTIS (2003). *Sodium channel beta4, a new disulfide-linked auxiliary subunit with similarity to beta2..* J Neurosci, 23(20):7577–7585.

[ZERANGUE et al. 1999] ZERANGUE, N., B. SCHWAPPACH, Y. N. JAN und L. Y. JAN (1999). *A new ER trafficking signal regulates the subunit stoichiometry of plasma membrane K(ATP) channels..* Neuron, 22(3):537–548.