

PUBLICATION LIST

BOOK CHAPTERS

10. Cornelius N, Gregersen N, **Tümer Z**, Olsen RKJ (2014) Oxidative Stress and Mitochondrial Defect in Neuromuscular Disorders: Source or Symptoms? In: Oxidative Stress: Causes, Role in Diseases and Biological Effects. Nova Science Publishers, Inc., New York, USA (in press).
9. Brender McNair J, McNair P, Dennis L, **Tümer Z** (2013) ATCG - An Applied Theory of Clinical Genetics. In: The Mereon Matrix: Unity, Perspective and Paradox (eds. Dennis L, Brender McNair J, Kauffman LH), Elsevier, Amsterdam, Netherlands, pp 347-516.
8. Mechior L, **Tümer Z** (2011) Genetics of Tourette Disease. In: Tourette Disease (eds. Von Plessen K, Thomsen PH), Dansk Psykologisk Forlag (in danish).
7. **Tümer Z**, Horn N (2004) Menkes disease. In: Neurocutaneous Syndromes (eds. Roach ES, Miller VS), Cambridge University Press, pp 222-233.
6. **Tümer Z**, Horn N, Jensen LR (2002) Expression profiling in Menkes disease. In: Handbook of Copper Pharmacology and Toxicology (ed. Massaro EJ), Humana Press, Totowa, New Jersey, pp 343-356.
5. Horn N, **Tümer Z** (2002) Menkes disease and the occipital horn syndrome. In: Connective Tissue and Its Heritable Disorders: Molecular, Genetic, and Medical Aspects (eds. Royce PM, Steinmann B), John Wiley and Sons Inc., New York, 2nd edition, pp 651-685.
4. Cox D, **Tümer Z** Roberts EA (2000) Disorders of Copper Metabolism. In: Inborn Metabolic Diseases (eds. Fernandes J, Saudubray J, van den Berghe G), Springer-Verlag, Berlin, 3rd edition, pp 385-391.
3. **Tümer Z**, Horn N (1999) Molecular genetics of Menkes disease. In: Metals and Genetics (ed. Sarkar B), Kluwer Academic / Plenum Publishers, New York, Boston, Dordrecht, London, Moscow, 2nd edition, pp 279-290.
2. **Tümer Z**, Horn N (1999) Hereditary disorders of copper metabolism. In: Genetic aberrancies and neurodegenerative disorders (ed. Mattson MP), Advances in Cell Aging and Gerontology, Jai Press Inc., Vol 3; pp 355-391.
1. Horn N, Tønnesen T, **Tümer Z** (1995) Variability in Clinical Expression of an X-linked Copper Disturbance, Menkes Disease. In: Metals and Genetics (ed. Sarkar B), Marcel Dekker Inc., pp 285-303.

MANUSCRIPTS

142. Sarri C, Douzgou S, Kontos H, Anagnostopoulou K, **Tümer Z**, Grigoriadou M, Petersen MB, Kokotas H, Merou K, Pandelia E, Giouroukou E, Papanikolaou K, Côté GB, Gyftodimou Y. The array-CGH characterization and 35-year follow up report of a patient with a ring chromosome 2. Review of the literature and reevaluation of the 'ring syndrome' phenotype. Cytogenetics and Genome Research (accepted).
141. Larsen CG, Gyldenløve M, Jønch AE, Charabi B, **Tümer Z**. A three-generation family with idiopathic facial palsy suggesting an autosomal dominant inheritance with high penetrance (2015) Otolaryngology (accepted).
140. Bertelsen B, Melchior L, Jensen LR, Groth C, Nazaryan L, Debes NM, Skov L, Xie G, Sun W, Brøndum-Nielsen K, Kuss AW, Chen W, **Tümer Z** (2014). A t(3;9)(q25.1;q34.3) translocation leading to OLFM1 fusion transcripts in Gilles de la Tourette syndrome, OCD and ADHD. Psychiatry Research (accepted).
139. Kashevarova AA, Nazarenko LP, Schultz-Pedersen S, Skryabin NA, Salyukova OA, Chechetkina NN, Tolmacheva EN, Rudko AA, Magini P, Graziano C, Romeo G, Joss S, **Tümer Z**, Lebedev IN (2014) Single gene microdeletions and microduplication of 3p26.3 in

- three unrelated families: CNTN6 as a new candidate gene for intellectual disability. *Mol Cytogenet* 7:391.
138. Chatron N, Haddad V, Andrieux J, Désir J, Boute O, Dieux A, Baumann B, Drunat S, Gérard M, Bonnet C, Leheup B, Till M, Rossi M, Flori E, Alembik Y, Stewart H, McParland J, Hall C, Bernardini L, Cast P, Roos L, **Tümer Z**, Fagan K, Hackett A, Bain N, van Haeringen A, Ruivekamp C, Benzacken B, Sanlaville D, Edery P, Aboura A, Schluth-Bolard C (2014) Genotype-phenotype correlation in 18 patients carrying a 1q24q25 deletion supports evidence for major contribution of DNMT3A locus to the phenotype. *Am J Med Genet A* (accepted).
 137. Boyle MI, Jespersgaard C, Brøndum-Nielsen K, Bisgaard A, **Tümer Z** (2014) Cornelia de Lange Syndrome. *Clin Genet Sep 11* [Epub ahead of print].
 136. Kaalund S, Venø MT, Bak M, Møller RS, Laursen H, Madsen F, Broholm H, Quistorff B, Ulldal P, Tommerup N, Kaupinen S, Sabers A, Fluiter K, Møller LB, Nossent AY, Silahatoglu A, Kjems J, Aronica E, **Tümer Z** (2014) miR-218 and miR-204 regulate the glutamatergic pathway in mesial temporal lobe epilepsy and hippocampus sclerosis. *Epilepsia* 55:2017-2027.
 135. De Rocker N, Vergult S, Jacobs E, Zeesman S, Bena F, Bockaert N, de Ravel T, Devriendt K, Giglio S, Faivre L, Joss S, Koolen D, Maas S, Bongers EM, Novara F, Nowaczyk MJM, Peeters H, Pohstra A, Poelens F, Rosenberg C, Thevenon J, **Tümer Z**, Vanhauwaert S, Varvagiannis K, Willaert A, Willemsen M, Willems M, Zuffardi O, Coucke P, Speleman F, Kleefstra T, Menten B (2014) Refinement of the critical 2p25.3 deletion region: the role of MYT1L in intellectual disability and obesity. *Genet Med Sep 18* [Epub ahead of print].
 134. Rasmussen MA, Holst B, **Tümer Z**, Johnsen MG, Zhou S, Stummann TC, Hyttel P, Clausen C (2014) Transient p53 suppression increases reprogramming of human fibroblasts without affecting apoptosis and DNA damage. *Stem Cell Reports* 9:403-413.
 133. Bertelsen B, Melchior L, Groth C, Debes NM, Skov L, Fagerlund B, Mikkelsen JD, **Tümer Z** (2014) Association of the CHRNA7 promoter variant -86T with Tourette syndrome and comorbid OCD. *Psychiatry Res* 219:710-711.
 132. Minocherhomji S, Hansen C, Kim H-G, Mang Y, Bak M, Guldborg P, Papadopoulos N, Eiberg H, Doh GD, Møllgård K, Hertz JM, Ropers H-H, **Tümer Z**, Tommerup N, Kalscheuer VM, Silahatoglu A (2014) Epigenetic remodeling and deregulation of SAPAP4/DLGAP4 is linked with early-onset cerebellar ataxia. *Hum Mol Genet* 23:6163-6176.
 131. Rajkumar A, Christensen J, Mattheisen M, Jacobsen I, Bache I, Pallesen J, Grove J, Qvist P, McQuillin A, Gurling H, **Tümer Z**, Mors O, Børglum A. P, Andrew G (2014) Analysis of t(9;17)(q33.2;q25.3) chromosomal breakpoint regions and genetic association reveals novel candidate genes for bipolar disorder. *Bipolar Disorders Jul 23* [Epub ahead of print].
 130. Delgado MS, Camprubi C, **Tümer Z**, Martienz F, Mila M, Monk D (2014) Screening individuals with Intellectual Disability, Autism and Tourette's syndrome for KCNK9 mutations and aberrant DNA methylation within the 8q24 imprinted cluster. *Am J Med Gen B – Neuropsychiatr Genet* 165:472-478.
 129. Grønskov K, Diness B, Stahlhut M, Zilmer M, **Tümer Z**, Bisgaard A-M, Brøndum-Nielsen K (2014) Mosaicism for c.431_454dup in ARX causes a mild Partington syndrome phenotype. *Eur J Med Genet* 57:284-287.
 128. Kwasny D, Mednova O, Vedarethinam I, Dimaki M, Silahatoglu A, **Tümer Z**, Almdal K, Svendsen WE (2014). A semi-closed device for metaphase chromosome spreading. *Micromachines* 5:158-170.
 127. Bertelsen B, Melchior L, Jensen LR, Groth C, Glenthøj B, Rizzo R, Debes NM, Skov L, Brøndum-Nielsen K, Paschou P, Silahatoglu A, **Tümer Z** (2014) Intragenic deletions affecting two alternative transcripts of the IMMP2L gene in patients with Tourette syndrome. *Eur J Hum Genet* 22:1283-1289.
 126. Zink A, Wohlleber E, Engels H, Rødningen O, Ravn K, Heilmann S, Rehnitz J, Katzorke N,

- Kraus C, Blichfeldt S, Hoffmann P, Reutter H, Brockschmidt F, Kreiß-Nachtsheim M, Vogt P, Prescott T, **Tümer Z**, Lee J (2014) Microdeletions including FMR1 in Three Female Patients with Intellectual Disability - further Delineation of the Phenotype and Expression Studies. *Molecular Syndromology* 5:65-75.
125. Møller RS, Jensen LR, Maas SM, Filmus J, Capurro M, Hansen C, Marcelis CLM, Ravn K, Andrieux J, Mathieu M, Kirchhoff M, Rødningen OK, de Leeuw N, Yntema HG, Froyen G, Vandewalle J, Ballon K, Klopocki E, Joss S, Tolmie J, Knecht AC, Lund AM, Hjalgrim H, Kuss AW, Tommerup N, Ullmann R, de Brouwer APM, Strømme P, Kjaergaard S, **Tümer Z***, Kleefstra T* (2014) X-linked congenital ptosis and associated intellectual disability, short stature, microcephaly, cleft palate, digital and genital abnormalities define novel Xq25q26 duplication syndrome. *Hum Genet* 133:625-638. * *Shared senior authorship, corresponding author*
124. Thevenon J, Callier P, Poquet H, Bache I, Menten B, Malan V, Cavaliere ML, Girod JP, Thauvin-Robinet C, El Chehadeh S, Pinoit JM, Huet F, Verges B, Petit JM, Mosca-Boidron AL, Marle N, Mugneret F, Masurel-Paulet A, Novelli A, **Tümer Z**, Loeys B, Lyonnet S, Faivre L (2014) 3q27.3 microdeletional syndrome: a recognisable clinical entity associating dysmorphic features, marfanoid habitus, intellectual disability and psychosis with mood disorder. *J Med Genet* 52:21-27.
123. Roos L, Fang M, Dali C, Jensen H, Christoffersen N, Wu B, Zhang J, Xu R, Harris P, Xu X, Grønskov K, **Tümer Z** (2013). A homozygous mutation in a consanguineous family consolidates the role of *ALDH1A3* in autosomal recessive microphthalmia. *Clin Genet* 86:276-281.
122. Bertelsen B, Debes NM, Hjermand LE, Skov L, Brøndum-Nielsen K, **Tümer Z** (2013) Chromosomal rearrangements in Tourette syndrome: Implications for identification of candidate susceptibility genes and review of the literature. *Neurogenetics* 14:197-203.
121. Melchior L, Bertelsen B, Debes NM, Groth C, Skov LL, Brøndum-Nielsen K, **Tümer Z** (2013) Microduplication of 15q13.3 and Xq21.31 in a family with Tourette Syndrome and comorbidities. *Am J Med Genet B Neuropsychiatr Genet* 162B:825-831.
120. Schönewolf-Greulich B, Ravn K, Hamborg-Petersen B, Brøndum-Nielsen K, **Tümer Z** (2013) Segregation of a 4p16.3 duplication with characteristic dysmorphic features, macrocephaly, speech delay and mild intellectual disability in a three generation family. *Am J Med Genet A* 161:2358-2362.
119. Gilling G, Rasmussen HB, Calloe K, Sequeira AF, Oliveira G, Almeida J, Lauritsen MB, Ullmann R, Boonen SE, Brøndum-Nielsen K, Kalscheuer V, **Tümer Z**, Vicente A, Schmitt N, Tommerup N (2013) Dysfunction of the heteromeric Kv7.3/Kv7.5 potassium channel is associated with autism spectrum disorders. *Front Genet* 4:54.
118. Sibbesen EC, Jespersgaard C, Alosi D, Bisgaard A-M, **Tümer Z** (2013) Ring chromosome 9 in a girl with developmental delay and dysmorphic features: case report and review. *Am J Med Genet A* 16:1447-1452.
117. Buttenschøn HN, Jacobsen IS, Grynderup MB, Hansen ÅM, Kolstad HA, Kaerlev L, Thomsen JF, Nordentoft M, Silahatoglu A, Tommerup T, **Tümer Z**, Krogh J, Børghlum AD, Mors O (2013) An association study between the norepinephrine transporter gene and depression. *Psych Genet* 23:217-221.
116. **Tümer Z** (2013) An overview and update of ATP7A mutations leading to Menkes disease and occipital horn syndrome. *Hum Mutat* 34:417-429.
115. Rendtorff ND, Schrijver I, Lodahl M, Rodriguez-Paris J, Johnsen T, Hansen EV, Nickelsen LAA, **Tümer Z**, Fagerheim T, Wetke R, Tranebjærg L (2013) SLC26A4 mutation frequency and spectrum in 109 Danish Pendred syndrome/DFNB4 probands and a report of nine novel mutations. *Clin Genet* 84:388-391.

114. Yasmeen S, Melchior L, Bertelsen B, Skov L, Debes NM, **Tümer Z** (2013) Sequence analysis of *SLITRK1* for var321 in Danish patients with Tourette syndrome and review of the literature. *Psych Genet* 23:130-133.
113. Boonen SE, Mackay DJG, Hahnemann JMD, Docherty L, Ellard S, Grønskov K, Lehmann A, Larsen LG, Haemers AP, Kockaerts Y, Dooms L, Vù DC, Ngoc CTB, Phuong NB, Kordonouri O, Sundberg F, Dayanikli P, Puthie V, Acerini C, Massoud AF, **Tümer Z**, Temple IK (2013). *ZFP57* and hypomethylation of multiple imprinted loci. *Diabetes Care* 36:505-512.
112. Brøgger AL, Kwasny D, Bosco FG, **Tümer Z**, Boisen A, Svendsen WE (2012). Centrifugally driven microfluidic disc for detection of chromosomal translocations. *Lab Chip* 12:4628-4638.
111. Jønch AE, Larsen LG, Pouplier S, Nielsen K, Brøndum-Nielsen K, **Tümer Z** (2012) Partial Duplication of 13q31.3-q34 and Deletion of 13q34 Associated with Diaphragmatic Hernia in a Fetus. *Am J Med Genet* 158A:2302-2308.
110. Becker K, Di Donato ND, Holder-Espinasse M, Andrieux J, Cuisset J-M, Vallée L, Plessis G, Jean N, Delobel B, Thuresson A-C, Annerén G, Ravn K, **Tümer Z**, Tinschert S, Schrock E, Jønch AE, Hackmann K (2012). De novo microdeletions of chromosome 6q14.1-q14.3 and 6q12.1-q14.1 in two patients with intellectual disability - further delineation of the 6q14 microdeletion syndrome and review of the literature. *Eur J Med Genet* 55:490-497.
109. Møller LB, Lenartowicz M, Zobot M-T, Josaine A, Burglen L, Bennett B, Riconda D, Fisher R, Janssens S, Mohammed S, Ausems M, **Tümer Z**, Horn N, Jensen TG (2012) Clinical expression of Menkes disease in females with normal karyotype. *Orphanet J Rare Dis* 7:6.
108. Kwasny D, Vedarethinam I, Shah P, Dimaki M, Silaharoglu A, **Tümer Z**, Svendsen WE (2012). Advanced microtechnologies for detection of chromosome abnormalities by fluorescent in situ hybridization. *Biomed Microdevices* 14:453-460.
107. **Tümer Z**, Bertelsen B, Gredal O, Magyari M, Nielsen KC, LuCamp, Grønskov K, Brøndum-Nielsen K (2012) A novel heterozygous nonsense mutation of the *OPTN* gene segregating in a Danish family with ALS. *Neurobiol Aging* 33:208.e1-5.
106. Boonen SE, Hahnemann JMD, Mackay D, Tommerup N, Brøndum-Nielsen K, **Tümer Z**, Grønskov K (2012) No evidence of pathogenic variants or maternal effect of ZFP57 as the cause of Beckwith-Wiedemann syndrome. *Eur J Hum Genet* 20:119-121.
105. Ravn K, Lindquist SG, Nielsen K, Dahm TL, **Tümer Z** (2012) Deletion of *CUL4B* leads to concordant phenotype in a monozygotic twin pair. *Clin Genet* 82:292-294.
104. Roos L, Grønskov K, Jensen H, **Tümer Z** (2012) Microphthalmia and Anophthalmia. *Ugeskrift for Læger* 174:713-716.
103. Bertelsen B, Melchior L, Skov LL, Debes NM, Brøndum-Nielsen K, **Tümer Z** (2012) Genetics of Tourette syndrom: actual knowledge and research. *Ugeskrift for Læger* 174:484-487.
102. Shah P, Vedarethinam I, Kwasny D, Andresen L, Skov S, Silaharoglu A, **Tümer Z**, Dimaki M, Svendsen W (2011) A novel integrated device for metaphase FISH sample preparation. *Micromachines* 2:116-128.
101. Schönewolf-Greulich B, Ronan A, Ravn K, Bækgaard P, Rendtoff N, Tranebjærg L, Brøndum-Nielsen K, **Tümer Z** (2011) Two new cases with microdeletion of 17q23.2 suggest presence of a candidate gene for sensorineural hearing loss within this region. *Am J Med Genet* 155A:2964-2969.
100. Kariminejad R, Lind-Thomsen A, **Tümer Z**, Erdogan F, Ropers HH, Tommerup N, Ullmann R, Møller RS (2011) High frequency of rare copy number variations in patients with structural brain malformations, intellectual disabilities and epilepsy. *Hum Mutat* 32:1427-1435.
99. Sarri C, Douzgou S, Gyftodimou J, **Tümer Z**, Ravn K, Pasparaki A, Sarafidou T, Kontos H, Kokotas H, Karadima G, Grigoriadou M, Pandelia E, Theodorou V, Moschonas NK, Petersen

- MB (2011) Complex Distal 10q Rearrangement in a Mildly Mentally Retarded Girl. Follow up of the Case and Review of the Literature of Non-acrocentric Satellited Chromosomes. *Am J Med Genet* 155A:2841-2854.
98. Ravn K, Roende G, Duno M, Fuglsang K, Eiklid KL, **Tümer Z**, Nielsen JB, Skjeldal OH (2011) Two new Rett syndrome families and review of the literature: expanding the knowledge of *MECP2* frameshift mutations. *Orphanet J Rare Dis* 6:58.
 97. Bertelsen B, **Tümer Z***, Ravn K* (2011) Three New Loci for Determining X Chromosome Inactivation Patterns. *J Mol Cytogenet* 13:537-540. (*Shared last authorship)
 96. Cingöz S, Bache I, Bjerglund L, Ropers HH, Tommerup N, Jensen H, Brøndum-Nielsen K, **Tümer Z** (2011). Interstitial Deletion of 14q24.3-q32.2 in a Male Patient with Plagiocephalus, BPES features, Developmental Delay and Congenital Heart Defects. *Am J Med Genet A* 155A:203-206.
 95. Gilling M, Ullmann R, Kristoffersson U, Møller M, Henriksen KF, Bugge M, Kalscheuer VM, Lundsteen C, **Tümer Z**, Tommerup N (2011). Biparental inheritance of chromosomal abnormalities in male twins with non-syndromic mental retardation. *Eur J Med Genet* 54:e383-388.
 94. **Tümer Z**, Klomp G (2011). Clinical utility gene card for: Menkes Disease. *Eur J Hum Genet* 19(10).
 93. Boonen SE, Hoffmann AL, Donnai D, **Tümer Z**, Ravn K (2011). Diploid/triploid mosaicism: A rare event or an under-diagnosed syndrome? *Eur J Med Genet* 54:374-375.
 92. Grønsvov K, Poole RL, Hahnemann JMD, Thomson J, **Tümer Z**, Temple IK, Murphy R, Ravn K, Melchior L, Dedic A, Dolmer B, Brøndum-Nielsen K, Boonen SE, Mackay D (2011). Deletions and rearrangements of the *h19/igf2* enhancer region in patients with Silver-Russell syndrome and growth retardation. *J Med Genet* 48:308-311.
 91. Nytofte NS, Serrano MA, Monte MJ, Gonzalez-Sanchez E, **Tümer Z**, Ladefoged K, Briz O, Marin JJG (2011). A homozygous nonsense mutation (c.214C>A) in biliverdin reductase alpha gene (*BLVRA*) results in accumulation of biliverdin during episodes of chole-stasis. *J Med Genet* 48:219-225.
 90. Skørring T*, **Tümer Z***, Møller LB (2011). Splice Site Mutations in the ATP7A Gene. *PLoS One* 6:e18599.
 89. Vedarethinam I, Shah P, Dimaki M, **Tümer Z**, Tommerup N, Svendsen WE (2010) Rapid Metaphase FISH on chip: Miniaturized microfluidic device for rapid in-situ hybridization. *Sensors* 10:9831-9846.
 88. Weisschuh N, De Baere E, Wissinger B, **Tümer Z** (2010). Clinical utility gene card for: Axenfeld Rieger syndrome. *Eur J Hum Genet* 19(3).
 87. Thienpont B, Zhang L, Postma AV, Breckpot J, Tranchevent L-J, van Loo P, Møllgård K, Tommerup N, Bache I, **Tümer Z**, van Engelen K, Menten B, Mortier G, Waggoner D, Gewillig M, Moreau Y, Devriendt K, Larsen LA (2010). Haplo-insufficiency of *TAB2* causes congenital heart defects in humans. *Am J Hum Genet* 86:839-849.
 86. Laszlo A, Endreffy E, **Tümer Z**, Horn N, Szabo J (2010). Molecular Genetic Mutation Analysis in Menkes Disease with Prenatal Diagnosis. *Clinical Neuroscience* 63:48-51.
 85. Sehested LT, Møller RS, Bache I, Andersen NB, Ullmann R, Tommerup N, **Tümer Z** (2010). Deletion of 7q34-q36.2 in patients with mental retardation, language delay, primary amenorrhea and dysmorphic features. *Am J Med Genet A* 152A:3115-3119.
 84. **Tümer Z**, Møller LB (2010). Menkes disease. *Eur J Hum Genet* 18:511-518.
 83. Roos L, Brøndum Nielsen K, **Tümer Z** (2009). A duplication Encompassing the *SHOX* Gene and the Downstream Evolutionarily Conserved Sequences. *Am J Med Genet* 149A:2900-2901.

82. Hilhorst-Hofstee Y*, **Tümer Z***, Born P, Knijnenburg J, Hansson K, Yatawara V, Stensbjerg J, Ullmann R, Arkesteijn G, Tommerup T, Larsen LA (2009). Molecular characterization of two patients with *de novo* interstitial deletions in 4q22-q24. *Am J Med Genet* 149A: 1830-1833. *Shared first authorship.
81. **Tümer Z**, Bach-Holm D (2009) Axenfeld-Rieger Syndrome and Spectrum of PITX2 and FOXC1 mutations *Eur J Hum Genet* 17:1527-1539.
80. Zhang L, **Tümer Z**, Møllgård K, Barbi G, Rossier E, Bendtsen E, Møller R, Ullmann R, He J, Papadopoulos N, Tommerup N, Larsen LA (2009). Characterization of a t(5;8)(q33;q22) translocation in a patient with mental retardation and congenital heart disease: implications for involvement of *RUNX1T1* in human brain and heart development *Eur J Hum Genet*. 17:1010-1018.
79. Brudzewsky D, Pedersen A, Claesson MH, Gad M, Kristensen NN, Lage K, Jensen T, Tommerup N, Larsen LA, Knudsen S, **Tümer Z** (2009). Genome-wide gene expression profiling of SCID mice with T-cell mediated colitis *Scan J Immun* 69:437-446.
78. Buysse K, Crepel A, Menten B, Pattyn F, Antonacci F, Veltman JA, Larsen LA, **Tümer Z**, de Klein A, van de Laar I, Devriendt K, Mortier G, Speleman F (2008). Mapping of 5q35 chromosomal rearrangements within a genomically unstable region. *J Med Genet* 45:672-678.
77. Erdogan F, Larsen LA, Zhang L, Tommerup Z, Chen W, Jacobsen JR, Schubert M, Jurkatis J, Tzschach A, Ropers HH, **Tümer Z**, Ullmann R (2008). High frequency of submicroscopic genomic aberrations detected by tiling path array comparative genome hybridisation in patients with isolated congenital heart disease. *J Med Genet* 45:704-709.
76. Kaalund S, Møller RS, Teszas A, Miranda M, Kosztolanyi G, Ullmann R, Tommerup N, **Tümer Z** (2008). Investigation of the chromosome 4 long arm deletion in two patients with a cryptic translocation and interstitial deletion using array CGH. *Am J Med Genet* 146A:2431-2434.
75. Møller RS, Hoeltzenbein M, Heye B, Kübart S, Vogel I, Ullmann R, Schröer A, Ruschmann J, Tommerup N, Ropers HH, **Tümer Z**, Kalscheuer VM (2008). Disruption of *DYRK1A* on chromosome 21q22.2 in two unrelated patients with microcephaly. *Am J Hum Genet* 82:1165-1170.
74. Engenheiro EL, Møller RS, Pinto M, Soares G, Nikanorova M, Marques I, Ullmann R, Tommerup N, **Tümer Z** (2008). Mowat-Wilson Syndrome: an underdiagnosed syndrome? *Clin Genet* 73:579-584.
73. Møller R, Schneider LM, Hansen CP, Bugge M, Ullmann R, Tommerup N, **Tümer Z** (2008). Balanced translocation in a patient with severe epilepsy and mental retardation disrupts the sodium channel gene *SCN1A*. *Epilepsia* 49:1091-1094.
72. Gilling M, Briciet-Lauritsen M, Møller M, Henriksen KF, Vicente A, Oliveira G, de Aguiar CC, Eiberg H, Andersen PS, Mors O, Brøndum-Nielsen K, Cotterill R, Lundsteen C, Ropers HH, Ullmann R, Bache I, **Tümer Z**, Tommerup N (2008). A 3.2Mb deletion on 18q12 in a patient with childhood autism and high-grade myopia. *Eur J Hum Genet* 16:312-319.
71. Vestergaard J, Lind-Thomsen A, Pedersen MW, Jarmer HO, Bak M, Hasholt L, Tommerup N, **Tümer Z**, Larsen LA (2008). *GLI1* is involved in cell cycle regulation and proliferation of NT2 embryonal carcinoma stem cells. *DNA Cell Biol* 27:251-256.
70. Erdogan F, Belloso JM, Gabau E, Ajbro KD, Guitart M, Ropers HH, Tommerup N, Ullmann R, **Tümer Z**, Larsen LA (2008). Fine mapping of a *de novo* interstitial 10q22-q23 duplication in a patient with congenital heart disease and microcephaly. *Eur J Med Genet* 51:81-86.
69. Hansen CP, Møller R, Tümer Z, Tommerup N (2007) The Genetic Basis of Epilepsy. The Danish Epilepsy Society. *Ugeskr Læger* 169:1102.
68. Møller RS, Hansen CP, Jackson GD, Ullmann R, Ropers HH, Tommerup N, **Tümer Z** (2007).

- Interstitial deletion of chromosome 4p associated with mild mental retardation, epilepsy and polymicrogyria of the left temporal lobe. *Clin Genet* 72:593-598.
67. Engenheiro E, Saravia J, Carreira I, Ramos L, Ropers HH, Silva E, Tommerup N, **Tümer Z** (2007). Cytogenetically invisible microdeletions involving PITX2 in Rieger syndrome. *Clin Genet* 72:464-470.
 66. Fickelscher I, Liehr T, Watts K, Barber JCK, Seibert R, Hertz JM, **Tümer Z**, Thomas NS (2007) Origins of the inv(2)(p11.2q13). *Am J Hum Genet* 81:847-856.
 65. Lage K, Karlberg EO, Størling ZM, Ólason PI, Pedersen AG, Rigina O, Hinsby AM, **Tümer Z**, Moreau Y, Pociot F, Tommerup N, Brunak S (2007) A human phenome-interactome network of protein complexes implicated in genetic disorders. *Nat Biotech* 25:309-316.
 64. Belloso JM, Bache I, Guitart M, Caballin MR, Halgren C, Kirchhoff M, Ropers HH, Tommerup N, **Tümer Z** (2007) Disruption of the *CNTNAP2* gene in a t(7;15) translocation family without symptoms of Gilles de la Tourette syndrome. *Eur J Hum Genet* 15:711-713.
 63. Jakobsen LP, Ullmann R, Christensen SB, Jensen KE, Mølsted K, Henriksen KF, Hansen C, Knudsen MA, Larsen LA, Tommerup N, **Tümer Z** (2007). Pierre Robin Sequence may be caused by dysregulation of both *SOX9* and *KCNJ2*. *J Med Genet* 44:381-386.
 62. Zhang L, **Tümer Z**, Jacobsen JR, Andersen PS, Tommerup N, Larsen LA. Screening of 99 Danish patients with congenital heart disease for *GATA4* mutations (2006). *Genetic Testing* 10:277-280.
 61. Bisgaard A-M, Kirchhoff M, **Tümer Z**, Skovby F, Jepsen B, Brøndum-Nielsen K, Cohen M, Hamborg-Petersen B, Tommerup N, Bryndorf T (2006). Additional Chromosomal Abnormalities in Patients with a Previously Detected Abnormal Karyotype, Mental Retardation, and Dysmorphic Features. *Am J Med Genet A* 140:2180-2187.
 60. Cingöz S, Bisgaard AM, Bache I, Bryndorf T, Kirchhoff M, Petersen, W, Ropers H-H, Maas N, Van Buggenhout G, Tommerup N, **Tümer Z** (2006) 4q35 deletion and 10p15 duplication associated with immunodeficiency *Am J Med Genet A* 140:2231-2235.
 59. Gilling M, Düllinger J, Gesk S, Metzke-Heidemann, Siebert R, Meyer T, Brøndum-Nielsen K, Tommerup N, Ropers H-H, **Tümer Z**, Kalscheuer V, Thomas NS (2006). Breakpoint cloning and haplotype analysis indicate a single origin of the common inv(10)(p11.2q21.2) among Northern Europeans. *Am J Hum Genet* 78:878-883.
 58. Vestergaard J, Pedersen MW, Pedersen N, Ensinger C, **Tümer Z**, Tommerup N, Poulsen HS, Larsen LA (2006). Hedgehog signaling in small-cell lung cancer: frequent *in vivo* but a rare event *in vitro*. *Lung Cancer* 52:281-290.
 57. Bækvad-Hansen M, **Tümer Z**, Delicado A, Erdogan F, Tommerup N, Larsen LA (2006). A 2.2 Mb microdeletion in 5q35 associated with microcephaly and congenital heart disease *Am J Med Genet* 140A:427-433.
 56. Długaszewska B, Silahatoglu A, Menzel C, Kübart S, Cohen M, Mundlos S, **Tümer Z**, Kjaer K, Frederich U, Ropers H-H, Tommerup N, Neitzel H, Kalscheuer V (2006). Breakpoints around the HOXD cluster result in different Limb Malformations. *J Med Genet* 2:111-118.
 55. Møller LB, Bukrinsky J, **Tümer Z**, Mølgaard A, Larsen S, Horn N (2005). Identification and analysis of 21 novel disease causing amino acid substitutions in the conserved part of ATP7A. *Hum Mutat* 26:1-10.
 54. Sogaard M, **Tümer Z**, Hjalgrim H, Hahnemann J, Friis B, Ledaal P, Pedersen VF, Baekgaard P, Tommerup N, Cingöz S, Duno M, Brøndum-Nielsen K (2005). Subtelomeric study of 132 patients with mental retardation reveals 9 chromosomal anomalies and contributes to the delineation of phenotypes of deletions of 1pter, 2qter, 4pter, 5qter and 9qter. *BMC Med Genet* 6:21.
 53. **Tümer Z**, Henriksen AM, Bache I, Larsen LA, Brixen K, Illum N, Rasmussen K, Tommerup N

- (2005) The eponymous Jacobsen syndrome: Mapping the breakpoints of the original family suggests an association between the distal 1.1 Mb of chromosome 21 and osteoporosis in Down syndrome. *Am J Med Genet* 135A:339-341.
52. Boonen SE, Stahl D, Rosenberg T, Kreiborg S, Kalscheuer V, Larsen LA, Tommerup N, Brøndum-Nielsen K, **Tümer Z** (2005) Delineation of an interstitial 9q22 deletion in basal cell nevus syndrome. *Am J Med Genet* 132A:324-328.
 51. Poulsen L, Møller LB, Plunkett K, Belmont J, **Tümer Z**, Horn N (2004). X-Linked Menkes Disease: First Documented Report of Germline Mosaicism. *Genet Test* 8:286-291.
 50. Henriksen AM, Tranebjerg L, **Tümer Z**, Tommerup N, Larsen LA (2004) Identification of a novel *EYAI* splice-site mutation in a Danish branchio-oto-renal syndrome family. *Genetic Test* 8:404-406.
 49. Bache I, Van Assche E, Cingöz S, Bugge M, **Tümer Z**, Hjorth M, Skovgaard LT, Lundsteen C, Niebuhr E, Fenger K, Lespinasse J, Liebaers I, Bonduelle M, Tournaye H, Tommerup N, *MCNdb* centers (2004) An excess of chromosome 1 breakpoints in male infertility. *Eur J Hum Genet* 12:993-1000.
 48. **Tümer Z**, Harboe TL, Blennow E, RH, Max Planck, Tommerup N, Brøndum-Nielsen K (2004) Molecular cytogenetic characterization of ring chromosome 15 in three unrelated patients. *Am J Med Genet* 130A:340-344.
 47. Hertz JM, Sivertsen B, Silaharoglu A, Bugge M, Kalscheuer V, Weber A, Wirth J, Ropers RR, Tommerup N, **Tümer Z** (2004) Early-onset, non-progressive and mild cerebellar ataxia co-segregating with a familial balanced translocation t(8;20)(p22;q13). *J Med Genet* 41:E25.
 46. Silaharoglu AN, Jensen LR, Harboe TL, Hansen C, Horn P, Bendixen C, Tommerup N, **Tümer Z** (2004) Sequencing and mapping of the porcine *CCS* gene. *Animal Genet* 35: 353-354.
 45. Midro AT, Panasiuk B, **Tümer Z**, Stankiewicz P, Silaharoglu A, Lupski JR, Zemanova Z, Stasiewicz-Jarocka B, Hubert E, Tarasow E, Famulski W, Zadrozna-Totwińska B, Wasilewska E, Kirchhoff M, Kalscheuer V, Michalova K, Tommerup N (2004) Interstitial deletion 9q22.32-q33.2 associated with additional familial translocation t(9;17)(q34.11;p11.2) in a patient with Gorlin-Goltz syndrome and features of nail-patella syndrome. *Am J Med Genet* 124A:179-191.
 44. **Tümer Z**, Møller LB, Horn N (2003) Screening of 383 unrelated patients affected with Menkes disease and finding of 57 gross deletions in the *ATP7A* gene. *Human Mutation* 22:457-464.
 43. Harboe TL, Jensen LR, Hansen C, Horn P, Bendixen C, Tommerup N, **Tümer Z** (2003) Cloning, characterization and chromosomal localisation of the *Sus scrofa SLC11A1* gene. *Animal Genet* 34:1-3.
 42. Poulsen L, Horn N, Heilstrup H, Lund C, **Tümer Z**, Møller LBM (2002) X-linked Recessive Menkes Disease: Identification of Partial Gene Deletions in Affected Males. *Clin Genet* 62:449-457.
 41. Harboe TL, Tommerup N, **Tümer Z** (2002) Assignment of *mZ13* to murine chromosome 4 with radiation hybrid mapping *Cytogenet Genome Res* 97:276A.
 40. Silaharoglu AN, Brøndum-Nielsen K, Gredal O, Werdelin L, Panas M, Petersen MB, Tommerup T, **Tümer Z** (2002) Human *CCS* gene: Genomic organization and exclusion as a candidate for amyotrophic lateral sclerosis (ALS). *BMC Genetics* 3:5.
 39. **Tümer Z**, Croucher PJP, Jensen LR, Hampe J, Hansen C, Kalscheuer V, Ropers HH, Tommerup N, Schreiber S (2002) Genomic structure, chromosome mapping and expression analysis of the human *AVIL* gene, and its exclusion as a candidate for IBD2. *Gene* 288:179-185.
 38. Kirchhoff M, Rose H, Maahr J, Gerdes T, Lespinasse J, Bugge M, Tommerup N, **Tümer Z**, Lespinasse J, Jensen PKA, Wirth J, Lundsteen C (2000) High resolution comparative genomic hybridization analysis reveals imbalances in dyschromosomal patients with normal or

- apparently balanced conventional karyotypes. *Eur J Hum Genet* 8:661-668.
37. Rendtorff ND, Vissing H, **Tümer Z**, Silahtaroglu A, Tommerup N (2000) Assignment of the NR2E3 gene to mouse chromosome 9 and to human chromosome 15q22.33-->q23. *Cytogenet Cell Genet.* 89:279-280.
 36. Harboe TL, **Tümer Z**, Hansen C, Jensen NA, Tommerup T (2000) Assignment of the human zinc finger gene, ZNF288, to chromosome 3 band q13.2 by radiation hybrid mapping and fluorescence in situ hybridization. *Cytogenet Cell Genet* 89:156-157.
 35. Møller LB, **Tümer Z**, Lund C, Petersen C, Cole T, Hanusch R, Seidel J, Jensen LR, Horn N (2000) Similar splicing mutations of the ATPA gene lead to different phenotypes: Classical Menkes disease or the occipital horn syndrome. *Am J Hum Genet* 66:1211-1220.
 34. Horn N, **Tümer Z** (1999) Molecular genetics of intracellular copper transport (1999) *J Trace Elem Exp Med* 12:297-313 (invited review).
 33. **Tümer Z**, Møller LB, Horn N (1999) Mutation spectrum of *ATP7A*, the gene defective in Menkes disease. *Adv Exp Med Biol.* 448:83-96 (Invited review).
 32. Jensen PY, Bonander N, Horn N, **Tümer Z**, Farver O (1999) Expression, purification and copper binding studies of the first metal binding domain of Menkes protein. *Eur J Biochem* 264: 890-896.
 31. Silahtaroglu A, Hol F, Jensen PKA, Erdel M, Duba H, Geurds MPA, Knoers NVAM, Mariman ECM, **Tümer Z**, Uttermann G, Wirth J, Bugge M, Tommerup N (1999) Molecular cytogenetic detection of 9q34-breakpoints associated with Nail-Patella syndrome. *Eur J Hum Genet* 7: 68-76.
 30. Jankov RP, Boerkoel CF, Hellmann J, Sirkin WL, Al-Maghrabi J, **Tümer Z**, Horn N, Feigenbaum A (1998) Lethal neonatal Menkes disease with severe vasculopathy and fractures. *Acta Paediatrica* 87:1297-1300.
 29. **Tümer Z**, Tommerup N, Binkert F, Back E, Brøndum-Nielsen, K (1998) Prader-Willi like phenotype and the proximal long arm of the X chromosome. *Am J Med Genet* 80:300-301.
 28. **Tümer Z** and Horn N (1998) Menkes disease: Underlying genetic defect and new diagnostic possibilities. *J Inher Metab Dis* 21:604-612 (Invited review).
 27. Jensen PY, Bonander N, Karlsson BG, Horn N, **Tümer Z**, Farver O (1998) Investigation of the copper-binding sites, in the Menkes disease protein, *ATP7A*. *J Inher Metab Dis* 21:195-198.
 26. **Tümer Z** (1998) Genetics of Menkes disease. *J Trace Elem Exp Med* 11: 147-161 (Invited review).
 25. Silahtaroglu A, Güven SG, Hacıhanefioglu S, Cenani A, Wirth J, Tommerup N, **Tümer Z** (1998) Not para- nor peri-, but centric inversion of chromosome 12. *J Med Genet* 35:682-684.
 24. Kaler SG, **Tümer Z** (1998) Prenatal diagnosis of Menkes disease. *Prenat Diagn* 18:287-289 (Invited Commentary).
 23. Fresko I, Soy M, Hamuryudan V, Yurdakul S, **Tümer Z**, Yazici H (1998) Genetic anticipation in Behcet's syndrome *Ann Rheum Dis* 57:45-48.
 22. Christodoulou J, Danks DM, Sarkar B, Baerlocher K, Horn N, Casey R, **Tümer Z**, Clarke JTR (1998) Early treatment of Menkes disease with parenteral copper-histidine: an update of four treated patients. *Am J Med Genet* 76: 154-164.
 21. Vural B, Poda M, Atlioglu E, Kolusayin Ö, Cenani A, Morling N, **Tümer Z** (1998) Turkish population data on the short tandem repeat (STR) locus TPOX. *Int J Legal Med* 111:105-106.
 20. **Tümer Z**, Wolff D, Silahtaroglu A, Willard HF, Ørum A, Brøndum-Nielsen K (1998) Characterization of the supernumerary small marker X chromosome in two females with similar phenotypes. *Am J Med Genet* 76:45-50.
 19. **Tümer Z**, Horn N (1997) Menkes disease: Recent advances and new aspects. *J Med Genet* 34:265-274 (Invited review).

18. **Tümer Z**, Horn N (1997) The hereditary disorders of copper metabolism: Identification of the Menkes disease gene and subsequent advances. In: Cavdar AO (ed) Trace Elements in Humans, pp. 35-56. Proceedings of the Trace Elements Symposium, 9th Meeting of the Mediterranean Blood Club, 9-11 September 1995, Cappadocia, Turkey (Invited review).
17. **Tümer Z**, Lund CL, Vural B, Tolshave J, Tønnesen T, Horn N (1997) Identification of point mutations in 41 unrelated Menkes disease patients. *Am J Hum Genetics* 60:63-71.
16. **Tümer Z**, Horn N, Christodoulou J, Tønnesen T, Clarke JTR, Sarkar B (1996) Early copper histidine treatment for Menkes disease. *Nature Genetics* 12:11-13.
15. **Tümer Z**, Horn N (1996) Menkes disease: Recent advances and new insights into copper metabolism *Ann Med* 28:121-128.
14. **Tümer Z**, Vural B, Tønnesen T, Chelly J, Monaco AP, Horn N (1995) Characterization of the exon structure of the Menkes Disease Gene Using Vectorette PCR. *Genomics* 26:437-442.
13. Silahtaroglu AN, Hacıhanefioglu S, Yilmaz S, Tarkan Y, Cenani A, **Tümer Z** (1995) A small supernumerary marker chromosome X identified by in situ hybridization. *Clin Genet* 47:270-273.
12. **Tümer Z**, Berg A and Mikkelsen M (1995) Analysis of a whole arm translocation between chromosomes 18 and 20 using fluorescence in situ hybridization: Detection of a break and a partial deletion in the centromeric a-satellite sequences. *Hum Genet* 95:299-302.
11. Beck J, Enders E, Schliephacke M, Buchwald-Saal M, **Tümer Z** (1994) X;1 translocation in a female Menkes patient: Characterization by fluorescence in situ hybridization. *Clin Genet* 46:295-298.
10. George AM, Reed V, Chelly J, **Tümer Z**, Chartier F, Brown SDM, Horn N, Monaco AP, Boyd Y (1994) Physical linkage of the murine homologue of Menkes' syndrome (*Mnk*) and phosphoglycerate kinase (*Pgk-1*) analysis of *Mnk* in mottled mutants using PFGE. *Genomics* 22:27-35.
9. **Tümer Z**, Tønnesen T, Böhmman J, Marg W, Horn N (1994) First Trimester Diagnosis of Menkes Disease with DNA Analysis. *J Med Genet* 31:615-617.
8. **Tümer Z**, Tønnesen T, Horn N (1994) Detection of mutations in Menkes disease and implications in carrier diagnosis. *J Inher Metab Dis* 17:267-270.
7. Silahtaroglu A, **Tümer Z**, Kristensen T, Sottrup-Jensen L, Tommerup N (1993) Assignment of PAPPa to 9q33.1 by fluorescence in situ hybridization to mitotic and meiotic chromosomes. *Cytogenet Cell Genet* 62:214-216.
6. Kreuder J, Otten A, Fuder H, **Tümer Z**, Tønnesen T, Horn N, Dralle D (1993) Biochemical and clinical consequences of copper-histidine therapy in Menkes disease. *Europ J Ped* 152:828-832.
5. Tommerup N, **Tümer Z**, Tønnesen T, Horn N (1993) A cytogenetic survey in Menkes disease: Implications of chromosomal rearrangements in X-linked disorders. *J Med Genet* 30:314-315.
4. **Chelly J, Tümer Z***, Tønnesen T, Petterson A, Ishikawa-Brush Y, Tommerup N, Horn N, Monaco AP (1993) Isolation of a candidate gene for Menkes disease that encodes for a potential heavy metal binding protein. *Nature Genetics* 3:14-19. * **Joint first authorship.**
3. **Tümer Z**, Chelly J, Tommerup T, Ishikawa-Brush Y, Tønnesen T, Monaco AP, Horn N (1992) Characterization of a 1.0 Mb YAC contig spanning two chromosome breakpoints related to Menkes disease. *Hum Mol Genet* 1:483-489.
2. Horn N, Tønnesen T, **Tümer Z** (1992) Menkes Disease: An X-linked Neurological Disorder of the Copper Metabolism. *Brain Pathology* 2:351-362 (**cover theme**).
1. **Tümer Z**, Horn N, Tønnesen T, Tommerup N (1992) Mapping of the Menkes Locus to Xq13.3 distal to the X-inactivation center by an intrachromosomal insertion of the segment Xq13.3-q21.2. *Hum Genet* 88:668-672.

OTHER SCIENTIFIC PUBLICATIONS

1. **Tümer Z**, Bach-Holm D. Axenfeld-Rieger Syndrome. Orphanet (The portal for rare diseases and orphan drugs) Encyclopedia for Professionals. July 2011 (http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=782).
2. **Tümer Z**, Møller LBM. Menkes disease. Orphanet (The portal for rare diseases and orphan drugs) Encyclopedia for Professionals. May 2011 (http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=565).
3. **Tümer Z**, Møller LBM. Occipital horn syndrome. Orphanet (The portal for rare diseases and orphan drugs) Encyclopedia for Professionals. May 2011 (http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=198).
4. Book review by Zeynep **Tümer**: Emery and Rimoin's Principles and Practice of Medical Genetics. 5th edition. Volume 1-3. Rimoin DL, Connor JM, Pyeritz RE et al. (red). Philadelphia: Churchill Livingstone Elsevier, 2007, Ugeskr Læger 169:2651.
5. **Tümer Z**, Horn N, Tonnesen T, Christodoulou J, Clarke JTR, Sarkar B (2004) Two ATP7A mutations in Menkes Disease (In Human Gene Mutations – Gene Symbol ATP7A) Hum Genet 114:606-609.
6. **Tümer Z** (1996) Fine localisation of the defective gene in Menkes disease, an X-linked recessive disorder of copper metabolism. Danish Medical Bulletin 43:296-297.
7. Freud for Beginners, Richard Appignanesi, 1979, Writers and Readers Publishing Inc. Translated from English to Turkish by Zeynep **Tümer** and Yankı Yazgan, Karum Publications, 1991, Istanbul, Turkey.
8. Go'dag Danmark: En håndbog for udlændinge, indvandrere og flygtninge i Danmark (Hello Denmark: A handbook for foreigners, immigrants and refugees in Denmark). 1993, Stout publications, Herning, Denmark (Zeynep **Tümer** is member of the initiative group).

DATABASES

Curator of Menkes Disease and ATPase, Cu⁺⁺ transporting, alpha polypeptide (ATP7A), LOVDdb (Leiden Open Variation Database) https://grenada.lumc.nl/LOVD2/MD/home.php?select_db=ATP7A (2012).