REFERENCES

Adams, D.J., Biggs, P.J., Cox, T., Davies, R., van der Weyden, L., Jonkers, J., Smith, J., Plumb, B., Taylor, R., Nishijima, I., Yu, Y., Rogers, J., Bradley, A. (2004). "Mutagenic Insertion and Chromosome Engineering Resource (MICER)." Nature Genetics **36**(8): 867-871.

Adams, D.J., Quail, M.A., Cox, T., van der Weyden, L., Gorick, B.D., Su, Q., Chan, W., Davies, R., Bonfield, J.K., Law, F., Humphray, S., Plumb, B., Liu, P., Rogers, J., Bradley, A. (2005). "A genome-wide, end-sequenced 129Sv BAC library resource for targeting vector construction." Genomics **86**(6): 753-758.

Ahlbom, B., Sidenvall, R., Anneren, G. (1996). "Deletion of chromosome 21 in a girl with congenital hypothyroidism and mild mental retardation." American Journal of Medical Genetics **64**: 501-505.

Aoki, M., Yamamoto, K., Ohyama, S., Yamamura, Y., Takenoshita, S., Sugano, K., Minamoto, T., Kitajima, M., Sugimura, H., Shimada, S. (2005). "A genetic variant in the gene encoding the stress 70 protein chaperone family member STCH is associated with gastric cancer in the Japanese population." Biochemical and Biophysical Research Communications **335**(2): 566-574.

Aoki, M., Yamamura, Y., Noshiro, H., Sakai, K., Yokota, J., Kohno, T., Tokino, T., Ishida, S., Ohyama, S., Ninomiya, I., Uesaka, K., Kitajima, M., Shimada, S., Matsuno, S., Yano, M., Hiratsuka, M., Sugimura, H., Itoh, F., Minamoto, T., Maehara, Y., Takenoshita, S., Aikou, T., Katai, H., Yoshimura, K., Takahashi, T., Akagi, K., Sairenji, M., Yamamoto, K., Sasazuki, T. (2005). "A full genome scan for gastric cancer." Journal of Medical Genetics **42**: 83-87.

Asnicar, M.A., Smith, D.P., Yang, D.D., Heiman, M.L., Fox, N., Chen, Y.F., Hsiung, H.M., Koster, A. (2001). "Absence of cocaine- and amphetamine-regulated transcript results in obesity in mice fed a high caloric diet." Endocrinology **142**(10): 4394-4400.

Baldini, A., Lindsay, E.A. (1994). "Mapping human YAC clones by fluorescence in situ hybridization using Ah-PCR from single yeast colonies." Methods in Molecular Biology **33**: 75-84.

Baujat, G., Cormier-Daire, V. (2007). "Sotos syndrome." Orphanet Journal of Rare Diseases **2**(1): 1-6.

Benjamini, Y., Yekutieli, D. (2001). "The control of the false discovery rate in multiple testing under dependency." The Annals of Statistics **29, No. 4**: 1165-1188.

Besson, V., Brault, V., Duchon, A., Togbe, D., Bizot, J. C., Quesniaux, V.F.J., Ryffel, B., Herault, Y. (2007). "Modeling the monosomy for the telomeric part of human chromosome 21 reveals haploinsufficient genes modulating the inflammatory and airway responses." Human Molecular Genetics **16(17)**: 2040-2052.

Bi, W., Ohyama, T., Nakamura, H., Yan, J., Visvanathan, J., Justice, M.J., Lupski, J.R. (2005). "Inactivation of Rai1 in mice recapitulates phenotypes observed in chromosome engineered mouse models for Smith-Magenis syndrome." Human Molecular Genetics **14**(8): 983-995.

Bosch-Comas, A., Lindsten, K., Gonzàlez-Duarte, R., Masucci, M.G., Marfany, G. (2006). "The ubiquitin-specific protease USP25 interacts with three sarcomeric proteins." Cellular and Molecular Life Sciences **63**: 723-734.

Bouchard, C., Tremblay, A., Despres, J.P., Nadeau, A., Lupien, P.J., Theriault, G., Dussault, J., Moorjani, S., Pinault, S., Fournier G. (1990). "The response to the long-term overfeeding in identical twins." New England Journal of Medicine **322**(21): 1477-1482.

Bradley A., Evans, M., Kaufman M.H., Robertson E. (1984). "Formation of germ-line chimaeras from embryo-derived teratocarcinoma cell lines." Nature **309**(5965): 255-256.

Brun, M.E., Ruault, M., Ventura, M., Roizes, G., De Sario, A. (2003). "Juxtacentromeric region of human chromosome 21: a boundary between centromeric heterochromatin and euchromatic chromosome arms." Gene **312**: 41-50.

Buiting, K. (2010). "Prader-Willi syndrome and Angelman syndrome." American Journal of Medical Genetics Part C: Seminars in Medical Genetics **154C**(3): 365-376.

Cattanach, B.M., Barr, J.A., Evans, E.P., Burtenshaw, M., Beechey, C.V., Leff, S.E., Brannan, C.I., Copeland, N.G., Jenkins, N.A., Jones, J. (1992). "A candidate mouse model for Prader-Willi syndrome which shows an absence of Snrpn expression." Nature Genetics **2**(4).

Cavailles, V., Dauvois, S., L'Horset, F., Lopez, G., Hoare, S., Kushner, P.J., Parker, M.J. (1995). "Nuclear factor RIP140 modulates transcriptional activation by the estrogen receptor." The EMBO Journal **14**(15): 3741-3751.

Cecconi, M., Forzano, F., Milani, D., Cavani, S., Baldo, C., Selicorni, A., Pantaleoni, C., Silengo, M., Ferrero, G.B., Scarano, G., Della Monica, M., Fischetto, R., Grammatico, P., Majore, S., Zampino, G., Memo, L., Cordisco, E.L., Neri, G., Pierluigi, M., Bricarelli, F.D., Grasso, M. Faravelli, F. (2005). "Mutation analysis of the NSD1 gene in a group of 59 patients with congenital overgrowth." American Journal of Medical Genetics Part A **134A**(3): 247-253.

Chen, L., Pik Wong, M., Kwong Cheung, L., Samaranayake, L.P., Baum, L., Samman, N. (2005). "Frequent allelic loss of 21q11.1~q21.1 region in advanced stage oral squamous cell carcinoma." Cancer Genetics and Cytogenetics **159**: 37-43.

Chettouh, Z., Croquette, M.F., Delobel, B., Gilgenkrants, S., Leonard,C., Maunoury, C., Prieur, M., Rethore, M.O., Sinet,' P.M., Chery, M., Delabar J.M. (1995). "Molecular mapping of 21 features associated with partial monosomy 21: involvement of the APP-SOD1 region." American Journal of Human Genetics **57(1)**: 62-71.

Claudio, J.O., Zhu, Y.X., Benn, S.J., Shukla, A.H., McGlade, C.J., Falcioni, N., Stewart, A.K. (2001). "HACS1 encodes a novel SH3-SAM adaptor protein diferentially expressed in normal and malignant hematopoietic cells." Oncogene **20**: 5373-5377.

Cliby, W., Ritland, S., Hartmann, R., Dodson, M., Hailing, K.C., Keeney, G., Podratz, K.C., Jenkins, R.B. (1993). "Human epithelial ovarian cancer allelotype." Cancer Research **53**: 2393-2398.

Cole, T.R.P., Hughes, H.E (1994). "Sotos syndrome: a study of the diagnostic criteria and natural history." Journal of Medical Genetics **31**: 20-32.

Corral, J., Lavenir, I., Impey ,H., Warren, A.J., Forster, A., Larson, T.A., Bell, S., McKenzie, A.N.J., King, G., Rabbitts, T.H. (1996). "An Mll-Af 9 fusion gene made by homologous recombination causes acute leukemia in chimeric mice: a method to create fusion oncogenes." Cell **85**: 853-861.

Davies, B.S.J., Barnes, R.H., Tu, Y., Ren, S., Andres, D.A., Spielmann, H.P., Lammerding, J., Wang, Y., Young, S.G., Fong, L.G. (2010). "An accumulation of non-farnesylated prelamin A causes cardiomyopathy but not progeria." Human Molecular Genetics **19**(13): 2682-2694.

Davies, B.S.J., Yang, S.H., Farber, E., Lee, R., Buck, S.B., Andres, D.A., Spielmann, H.P., Agnew, B.J., Tamanoi, F., Fong, L.G., Young, S.G. (2008). "Increasing the length of progerin's isoprenyl anchor does not worsen bone disease or survival in mice with Hutchinson-Gilford progeria syndrome." Journal of Lipid Research **50**(1): 126-134.

DiGeorge, A.M. (1968). "Congenital absence of the thymus and its immunologic consequences: concurrence with congenital hypoparathyroidism. White Plains, New York: March of Dimes-Birth Defects Foundation." Birth Defects **IV**(1): 116-121.

Ding, F., Prints, Y., Dhar, M.S., Johnson, D.K., Garnacho–Montero, C., Nicholls, R.D., Francke, U. (2005). "Lack of Pwcr1/MBII-85 snoRNA is critical for neonatal lethality in Prader–Willi syndrome mouse models." Mammalian Genome **16**(6): 424-431.

dos Santos Aguiar, S., de Jesus Girotto Zambaldi, L., dos Santos, A.M., Pinto Jr., W., Brandalise, S.R. (2007). "Comparative genomic hybridization analysis of abnormalities in chromosome 21 in childhood osteosarcoma." Cancer Genetics and Cytogenetics **175**: 35-40.

Douglas, J., Tatton-Brown, K., Coleman, K., Guerrero, S., Berg, J., Cole, T.R.P., FitzPatrick, D., Gillerot, Y., Hughes, H.E., Pilz, D., Raymond, F.L., Temple, I.K., Irrthum, A., Schouten, J.P., Rahman, N. (2005). "Partial NSD1 deletions cause 5% of Sotos syndrome and are readily identifiable by multiplex ligation dependent probe amplification." Journal of Medical Genetics **42**(9): 1-7.

Elsea, S.H., Girirajan, S. (2008). "Smith–Magenis syndrome." European Journal of Human Genetics **16**(4): 412-421.

Engelmann, M., Hädicke, J., Noack, J. (2011). "Testing declarative memory in laboratory rats and mice using the nonconditioned social discrimination procedure." Nature Protocols **6**(8): 1152-1162.

Evans, M.J., Kaufman, M.H. (1981). "Establishment in culture of pluripotential cells from mouse embryos." Nature **292**(5819): 154-156.

Faravelli, F. (2005). "NSD1 mutations in Sotos syndrome." American Journal of Medical Genetics Part C **137C**: 24-31.

Firth, H.V., Richards, S.M., Bevan, A.P., Clayton, S., Corpas, M., Rajan, D., van Vooren, S., Moreau, Y., Pettett, R.M., Carter N.P. (2009). "DECIPHER: Database of Chromosomal Imbalance and Phenotype in Humans Using Ensembl Resources." American Journal of Human Genetics **84**: 524-533.

Foell, J.L., Hesse, M., Volkmer, I., Schmiedel, B.J., Neumann, I., Staege, M.S. (2008). "Membrane-associated phospholipase A1 beta (LIPI) is an Ewing tumour-associated cancer/testis antigen." Pediatrian Blood Cancer **51**: 228-234.

Fong, L., Ng, J.K., Lammerding, J., Vickers, T.E., Meta, M., Coté, N., Gavino, B., Qiao, X., Chang, S.Y., Young, S.R., Yang, S.H., Stewart, C.L., Lee, R.T., Bennett, C.F., Bergo, M.O., Young, S.G. (2006). "Prelamin A and lamin A appear to be dispensable in the nuclear lamina." Journal of Clinical Investigation **116**(3): 743-752.

Frazer, K.A., Sheehan, J.B., Stokowski, R.P., Chen, X., Hosseini, R., Cheng, J.F, Fodor, S.P.A., Cox, D.R., Patil N. (2001). "Evolutionarily conserved sequences on human chromosome 21." Genome Research **11**: 1651–1659.

Gitler, A.D., Lu, M.M., Epstein, J.A. (2004). "PlexinD1 and semaphorin signaling are required in endothelial cells for cardiovascular development." Developmental Cell **7**(1): 107-116.

Groet, J., Ives, J.H., Jones, T.A., Danton, M., Flomen, R.H., Sheer, D., Hrascan, R., Pavelic, K., Nizetic, D. (2000). "Narrowing of the region of allelic loss in 21q11-21 in squamous non-small cell lung carcinoma and

cloning of a novel ubiquitin-specific protease gene from the deleted segment." Genes, Chromosomes & Cancer **27**: 153-161.

Gu, W., Zhang, F., Lupski, J.R (2008). "Mechanisms for human genomic rearrangements." Pathogenetics **1**(1):4: 1-17.

Guo, G., Wang, W., Bradley, A. (2004). "Mismatch repair genes identified using genetic screens in Blm-deficient embryonic stem cells." Nature **429**: 891-895.

Hasle, H., Haunstrup Clemmensen, I., Mikkelsen, M. (2000). "Risks of leukaemia and solid tumours in individuals with Down's syndrome." Lancet **355**: 165-169.

Hattori, M., Fujiyama, A., Taylor, T.D., Watanabe, H., Yada, T., Park, H.S., Toyoda, A., Ishii, K., Totoki, Y., Choi, D.K., Groner, Y., Soeda, E., Ohki, M., Takagi, T., Sakaki, Y., Taudien, S., Blechschmidt, K., Polley, A., Menzel, U., Delabar, J., Kumpf, K., Lehmann, R., Patterson, D., Reichwald, K., Rump, A., Schillhabel, M., Schudy, A., Zimmermann, W., Rosenthal, A., Kudoh, J., Schibuya, K., Kawasaki, K., Asakawa, S., Shintani, A., Sasaki, T., Nagamine, K., Mitsuyama, S., Antonarakis, S.E., Minoshima, S., Shimizu, N., Nordsiek, G., Hornischer, K., Brant, P., Scharfe M, Schon O, Desario A, Reichelt J, Kauer G, Blocker H, Ramser J, Beck A, Klages S, Hennig S, Riesselmann L, Dagand E, Haaf T, Wehrmeyer S, Borzym K, Gardiner, K., Nizetic, D., Francis, F., Lehrach, H., Reinhardt, R., Yaspo, M.L.; Chromosome 21 mapping and sequencing consortium (2000). "The DNA sequence of human chromosome 21." Nature **405**: 311-319.

Hegele, R.A., Cao, H., Harris, S.B., Zinman, B., Hanley, A.J., Anderson, C.M. (2000). "Genetic variation in LMNA modulates plasma leptin and indices of obesity in aboriginal Canadians." Physiological Genomics **3**: 39-44.

Hegele, R.A., Murray W.H., Young T.K. (2001). "Common genomic variation in LMNA modulates indexes of obesity in Inuit." Journal of Clinical Endocrinology & Metabolism **86**(6): 2747-2751.

Heim, K.C., White, K.A., Deng, D., Tomlinson, C.R., Moore, J.H, Freemantle, S.J., Spinella, M.J. (2007). "Selective repression of retinoic acid target

genes by RIP140 during induced tumor cell differentiation of pluripotent human embryonal carcinoma cells." Molecular Cancer **6:57**: 1-18.

Heitmann, B.L., Lissner, L., Sorensen, T.I.A, Bengtsson C. (1995). "Dietary fat intake and weight gain in women genetically predisposed for obesity." American Journal in Clinical Nutrition **61**.

Hiramatsu, T., Sonoda, H., Takanezawa, Y., Morikawa, R., Ishida, M., Kasahara, K., Sanai, Y., Taguchi, R., Aoki, J., Arai, H. (2003). "Biochemical and molecular characterization of two phosphatidic acid-selective phospholipase A1s, mPA-PLA1 and mPA-PLA1." The Journal of Biological Chemistry **278**(49): 49438-49447.

Holecki, M., Wiecek, A. (2010). "Relationship between body fat mass and bone metabolism." Polskie Archiwum Medycyny Wewnetrznej **120**(9): 361-367.

Hoogenraad, C.C., Koekkoek, B., Akhmanova, A., Krugers, H., Dortland, B., Miedema, M., van Alphen, A., Kistler, W.M., Jaegle, M., Koutsourakis, M., Van Camp, N., Verhoye, M., van der Linden, A., Kaverina, I., Grosveld, F., De Zeeuw, C.I., Galjart, N. (2002). "Targeted mutation of Cyln2 in the Williams syndrome critical region links CLIP-115 haploinsufficiency to neurodevelopmental abnormalities in mice." Nature Genetics **32**(1): 116-127.

Huang, N., vom Baur, E., Garnier, J.M., Lerouge, T., Vonesch, J.L., Lutz, Y., Chambon, P., Losson, R. (1998). "Two distinct nuclear receptor interaction domains in NSD1, a novel SET protein that exhibits characteristics of both corepressors and coactivators." European Molecular Biology Organisation Journal **17**(12): 3398–3412.

Hubbard, T.J., Aken, B.L., Beal, K., Ballester, B., Caccamo, M., Chen, Y., Clarke, L., Coates, G., Cunningham, F., Cutts, T., Down, T., Dyer, S.C., Fitzgerald, S., Fernandez-Banet, J., Graf, S., Haider, S., Hammond, M., Herrero, J., Holland, R., Howe, K., Howe, K., Johnson, N., Kahari, A., Keefe, D., Kokocinski, F., Kulesha, E., Lawson, D., Longden, I., Melsopp, C., Megy, K., Meidl, P., Ouverdin, B., Parker, A., Prlic, A., Rice, S., Rios, D., Schuster, M., Sealy, I., Severin, J., Slater, G., Smedley, D., Spudich, G., Trevanion, S., Vilella, A., Vogel, J., White, S., Wood, M., Cox, T., Curwen, V., Durbin, R., Fernandez-

Suarez, X.M., Flicek, P., Kasprzyk, A., Proctor, G., Searle, S., Smith, J., Ureta-Vidal, A,. Birney, E. (2007). "Ensembl 2007." Nucleic Acids Research **35**(D6): 10-17.

Iliopoulos, D., Sekerli, E., Vassiliou, G., Sidiropoulou, V., Topalidis, A., Dimopoulou, D., Voyiatzis, N. (2006). "Patau syndrome with a long survival (146 months): A clinical report and review of literature." American Journal of Medical Genetics Part A **140A**(1): 92-93.

Jenuwein, T., Laible, G., Dorn, R., Reuter, G. (1998). "SET domain proteins modulate chromatin domains in eu- and heterochromatin." Cellular and Molecular Life Sciences **54**: 80-93.

Jiang, Y.H., Armstrong, D., Albrecht, U., Atkins, C.M., Noebels, J.L., Eichele, G., Sweatt, D.J., Beaudet, A.L. (1998). "Mutation of the Angelman ubiquitin ligase in mice causes increased cytoplasmic p53 and deficits of contextual learning and long-term potentiation." Neuron **21**: 799-811.

Joosten, A.M.S., de Vos, S., van Opstal, D., Brandenburg H., Gaillard, J.L.J., Vermeij-Keers, C. (1996). "Full monosomy 21, prenatally diagnosed by fluorescent in situ hybrydyzation." Prenatal Diagnosis **17(3)**: 271-275.

Katzaki, E., Morin, G., Pollazzon, M., Papa, F.T., Buoni, S., Hayek, J., Andrieux, J., Lecerf, L., Popovici, C., Receveur, A., Mathieu-Dramard, M., Renieri, A., Mari, F., Philip, N. (2010). "Syndromic mental retardation with thrombocytopenia due to 21q22.11q22.12 deletion: Report of three patients." American Journal of Medical Genetics Part A **152A(7)**: 1711-1717.

Katzenstein, J.M., Oghalai, J.S., Tonini, R., Baker, D., Haymond, J., Caudle, S.E. (2009). "Neurocognitive functioning of a child with partial trisomy 6 and monosomy 21." Neurocase **15**(2): 97-100.

Kesler, S.R. (2007). "Turner Syndrome." Child and Adolescent Psychiatric Clinics of North America **16**(3): 709-722.

Kimber, W.L., Hsieh, P., Hirotsune, S., Yuva-Paylor, L., Sutherland, H.F., Chen, A., Ruiz-Lozano, P., Hoogstraten-Miller, S.L., Chien, K.R., Paylor, R., Scambler, P.J., Wynshaw-Boris, A. (1999). "Deletion of 150 kb in the minimal DiGeorge/velocardiofacial syndrome critical region in mouse." Human Molecular Genetics **8**(12): 229-2237.

Kogan, J.H., Frankland, P.W., Silva, A.J. (2000). "Long-term memory underlying hippocampus-dependent social recognition in mice." Hippocampus **10**(1): 47-56.

Kohno, T., Kawanishi, M., Matsuda, S., Ichikawa, H., Takada, M., Ohki, M., Yamamoto, T., Yokota, J. (1998). "Homozygous deletion and frequent allelic loss of the 21q11.1-q21.1 region including the ANA gene in human lung carcinoma." Genes, Chromosomes & Cancer **21**: 236-243.

Korenberg, J.R., Kalousek, D.K., Anneren, G., Pulst, S.M., Hall, J.G., Epstein, C.J., Cox, D.R. (1991). "Deletion of chromosome 21 and normal intelligence: molecular definition of the lesion." Human Genetics **87**: 112-118.

Kucera, G.T., Bortner, D.M., Rosenberg, M.P. (1996). "Overexpression of an Agouti cDNA in the skin of transgenic mice recapitulates dominant coat color phenotypes of spontaneous mutants." Developmental Biology **173**(162-173).

Kurotaki, N., Harada, N., Shimokawa, O., Miyake, N., Kawame, H., Uetake, K., Makita, Y., Kondoh, T., Ogata, T., Hasegawa, T., Nagai, T., Ozaki, T., Touyama, M., Shenhav, R., Ohashi, H., Medne, L., Shiihara, T., Ohtsu, S., Kato, Z., Okamoto, N., Nishimoto, J., Lev, D., Miyoshi, Y., Ishikiriyama, S., Sonoda, T., Sakazume, S., Fukushima, Y., Kurosawa, K., Cheng, J.F., Yoshiura, K., Ohta, T., Kishino, T., Niikawa, N., Matsumoto, N. (2003). "Fifty microdeletions among 112 cases of Sotos syndrome: Low copy repeats possibly mediate the common deletion." Human Mutation **22**(5): 378-387.

Kurotaki, N., Harada, N., Yoshiura, K., Sugano, S., Niikawa, N., Matsumoto, N. (2001). "Molecular characterization of NSD1, a human homologue of the mouse Nsd1 gene." Gene **279**: 197-204.

Kurotaki, N., Imaizumi, K., Harada, N., Masuno, M., Kondoh, T., Nagai, T., Ohashi, H., Naritomi, K., Tsukahara, M., Makita, Y., Sugimoto, T., Sonoda, T., Hasegawa, T., Chinen, Y.,Tomita, H., Kinoshita, A., Mizuguchi, T., Yoshiura, K., Ohta, T., Kishino, T., Fukushima, Y., Niikawa, N., Matsumoto, N. (2002). "Haploinsufficiency of NSD1 causes Sotos syndrome." Nature Genetics **30**(4): 365-366.

Leonardsson, G., Steel J.H., Christian M., Pocock, V., Milligan, S., Bell, J., So, P.W., Medina-Gomez, G., Vidal-Puig, A., White, R., Parker, M.G. (2004). "Nuclear receptor corepressor RIP140 regulates fat accumulation." Proceedings of the National Academy of Sciences **101**(22): 8437-8442.

Li, H.H., Roy, M., Kuscuoglu, U., Spencer, C.M., Halm, B., Harrison, K.C., Bayle, J.H., Splendore, A., Ding, F., Meltzer, L.A., Wright, E., Paylor, R., Deisseroth, K., Francke, U. (2009). "Induced chromosome deletions cause hypersociability and other features of Williams–Beuren syndrome in mice." EMBO Molecular Medicine **1**: 50-65.

Linday, E.A. (2001). "Chromosomal microdeletions: dissecting Del22q11 syndrome." Nature Reviews in Genetics **2**(11): 858-868.

Lindsay, E.A., Botta, A., Jurecic, V., Carattini-Rivera, S., Cheah, Y.C., Rosenblatt, H.M., Bradley, A., Baldini, A. (1999). "Congenital heart disease in mice deficient for the DiGeorge syndrome region." Nature **401**: 379-383.

Lindsay, E.A., Vitelli, F., Su, H., Morishima, M., Huynh, T., Pramparo, T., Jurecic, V., Ogunrinu, G., Sutherland, H.F., Scambler, P.J., Bradley, A., Baldini, A. (2001). "Tbx1 haploinsufficiency in the DiGeorge syndrome region causes aortic arch defects in mice." Nature **410**: 97-101.

Lindstrand, A., Malmgren, H., Sahlen, S., Schoumans, J., Nordgren, A., Ergander, U., Holm, E., Anderlid, B.M., Blennow, E. (2010). "Detailed molecular and clinical characterization of three patients with 21q deletions." Clinical Genetetics **77**: 145-154.

Liu, P., Zhang, H., McLellan, A., Vogel, H., Bradley, A. (1998). "Embryonic lethality and tumorigenesis caused by segmental aneuploidy on mouse chromosome 11." Genetics **150**: 1155-1168.

Livak, K.J., Schmittgen, T.D. (2001). "Analysis of relative gene expression data using real-time quantitative PCR and the 22DDCT method." Methods **25**: 402-408.

Luan, J., Browne, P.O., Harding, A.H., Halsall, D.J., O'Rahilly, S., Chatterjee, V.K.K, Wareham N.J. (2001). "Evidence for gene-nutrient interaction at the PPARg locus." Diabetes **50**: 686-689.

Luo, G., Santoro, I.M., McDaniel, L.D., Nishijima, I., Mills, M., Youssoufian, H., Vogel, H., Schultz, R.A., Bradley, A. (2000). "Cancer predisposition caused by elevated mitotic recombination in Bloom mice." Nature Genetics **26**: 424-429.

Lyle, R., Béna, F., Gagos, S., Gehrig, C., Lopez, G., Schinzel, A., Lespinasse, J., Bottani, A., Dahoun, S., Taine, L., Doco-Fenzy, M., Cornillet-Lefèbvre, P., Pelet, A., Lyonnet, S., Toutain, A., Colleaux, L., Horst, J., Kennerknecht, I., Wakamatsu, N., Descartes, M., Franklin, J.C., Florentin-Arar, L., Kitsiou, S., Yahya-Graison, E.A., Maher, C., Sinet, P.M., Delabar, J.M., Antonarakis, S.E. (2008). "Genotype–phenotype correlations in Down syndrome identified by array CGH in 30 cases of partial trisomy and partial monosomy chromosome 21." European Journal of Human Genetics **17(4)**: 454-466.

Mégarbané, A., Ravel, A., Mircher, C., Sturtz, F., Grattau, Y., Rethoré, M.O., Delabar, J.M., Mobley, W.C. (2009). "The 50th anniversary of the discovery of trisomy 21: The past, present, and future of research and treatment of Down syndrome." Genetics in Medicine **11**(9): 611-616.

Merscher, S., Funke, B., Epstein, J.A., Heyer, J., Puech, A., Lu, M.M., Xavier, R.J., Demay, M.B., Russell, R.G., Jore, B.S., Lopez, M., Pandita,R.K., Lia, M., Carrion, D., Xu, H., Schorle, H., Kobler, J.B., Scambler, P., Wynshaw-Boris, A., Skoultchi, A.I., Morrow, B.E., Kucherlapati, R. (2001). "TBX1 Is Responsible for Cardiovascular Defects in Velo-Cardio-Facial/DiGeorge Syndrome." Cell **104**: 619-629.

Mitelman, F., Mertens, F., Johansson, B. (1997). "A breakpoint map of recurrent chromosomal rearrangements in human neoplasia." Nature Genetics Special Issue: 417-474.

Mitsui, S., Osako, Y., Yokoi, F., Dang, M.T.,Yuri, K., Li, Y., Yamaguchi, N. (2009). "A mental retardation gene, motopsin/neurotrypsin/prss12, modulates hippocampal function and social interaction." European Journal of Neuroscience **30**(12): 2368-2378.

Mori, M.A., Lapunzina, P., Delicado, A., Nunez, G., Rodriguez, J.I., de Torres, M.L., Herrero, F., Valverde, E., Lopez-Pajares, I. (2004). "A prenatally diagnosed patient with full monosomy 21: Ultrasound, cytogenetic,

clinical, molecular, and necropsy findings." American Journal of Medical Genetics **127A**(1): 69-73.

Nagai, T., Matsumoto, N., Kurotaki, N., Harada, N., Niikawa, N., Ogata, T., Imaizumi, K.,Kurosawa, K., Kondoh, T., Ohashi, H., Tsukahara, M., Makita, Y., Sugimoto, T., Sonoda, T., Yokoyama, T., Uetake, K., Sakazume, S., Fukushima, Y., Naritomi, K. (2003). "Sotos syndrome and haploinsufficiency of NSD1: clinical features of intragenic mutations and submicroscopic deletions." Journal of Medical Genetics **40**: 285-289.

Nieters, A., Becker, N., Linseisen, J. (2002). "Polymorphisms in candidate obesity genes and their interaction with dietary intake of n-6 polyunsaturated fatty acids affect obesity risk in a sub-sample of the EPIC-Heidelberg cohort." European Journal of Nutrition **41**(5): 210-221.

O'Gorman, S., Dagenais, N.A., Qian, M., Marchuk, Y. (1997). "Protamine-Cre recombinase transgenes efficiently recombine target sequences in the male germ line of mice, but not in embryonic stem cells." Proceedings of the National Academy of Sciences of the United States of America **94**: 14602–14607.

Ohgaki, K., Iida, A., Kasumi, F., Sakamoto, G., Akimoto, M., Nakamura, Y., Emi, M. (1998). "Mapping of a new target region of allelic loss to a 6-cM interval at 21q21 in primary breast cancers." Genes, Chromosomes & Cancer(23): 244-247.

Olson, L.E., Richtsmeier, J.T., Leszl, J., Reeves, R.H. (2004). "A chromosome 21 critical region does not cause specific Down syndrome phenotypes." Science **306(5696)**: 687-690.

Olson, L.E., Roper, R.J., Baxter, L.L., Carlson, E.J., Epstein, C.J., Reeves, R.H. (2004). "Down syndrome mouse models Ts65Dn, Ts1Cje, and Ms1Cje/Ts65Dn exhibit variable severity of cerebellar phenotypes." Developmental Dynamics **230**(3): 581-589.

Olson, L.E., Roper, R.J., Sengstaken, C.L., Peterson, E.A., Aquino, V., Galdzicki, Z., Siarey, R., Pletnikov, M., Moran, T.H., Reeves, R.H. (2007). "Trisomy for the Down syndrome 'critical region' is necessary

but not sufficient for brain phenotypes of trisomic mice." Human Molecular Genetics **16(7)**: 774-4782.

Otterson, G.A., Flynn, G.C., Kratzkel, R.A., Coxon, A., Johnston, P.G., Kaye, F.J. (1994). "Stch encodes the 'ATPase core' of a microsomal stress70 protein." The EMBO Journal **13**(5): 1216-1225.

Overbeek, P.A., Aguilar-Cordova, E.,Hanten, G., Schaffner, D.L., Patel, P., Lebovitz, R.M., Lieberman, M.W. (1991). "Coinjection strategy for visual identification of transgenic mice." Transgenic Research **1**: 31-37.

Poehlman, E.T., Tremblay, A., Despres, J.P., Fontaine, E., Perusse, L., Theriault, G., Bouchard, C. (1986). "Genotype-controlled changes in body composition and fat morphology following overfeeding in twins." American Journal of Clinical Nutrition(43): 723-731.

Prader, A., Labhart, A., Willi, H. (1956). "Ein Syndrom von Adipositas, Kleinwuchs, Kryptorchismus und Oligophrenie nach Myatonieartigem Zustand im Neugeborenenalter." Schweizerische Medizinische Wochenschrift **86**: 1260-1261.

Puech, A., Saint-Jore, B., Merscher, S., Russell, R.G., Cherif, D., Sirotkin, H., Xu, H., Factor, S., Kucherlapati, R., Skoultchi, A.I. (2000). "Normal cardiovascular development in mice deficient for 16 genes in 550 kb of the velocardiofacialy DiGeorge syndrome region." Proceedings of the National Academy of Sciences **97**(18): 10090-10095.

Rabbitt, T.H. (1994). "Chromosomal translocations in human cancer." Nature **372**: 143-149.

Ramirez-Solis, R., Davis, A.C., Bradley, A. (1993). "Gene targeting in embryonic stem cells." Methods in Enzymology **225**: 855-878.

Ramirez-Solis, R., Liu, P., Bradley, A. (1995). "Chromosome engineering in mice." Nature **378**: 720-724.

Reymond, A., Marigo, V., Yaylaoglu, M.B., Leoni, A., Ucla, C., Scamuffa, N., Caccioppoli, C., Dermitzakis, E.T., Lyle, R., Banfi, S., Eichele, G., Antonarakis, S.E., Ballabio, A. (2002). "Human chromosome 21 gene expression atlas in the mouse." Nature **420**(6915): 582-586.

Richter, K., Wolf, G., Engelmann, M. (2005). "Social recognition memory requires two stages of protein synthesis in mice." Learning & Memory **12**(4): 407-413.

Riegel, M., Hargreaves. P., Baumer, A., Guc-Scekic, M., Ignjatovic, M., Schinzel A. (2005). "Unbalanced 18q/21q translocation in a patient previously reported as monosomy 21." European Journal of Medical Genetics **48(2)**: 167-174.

Roberson, E.D.O., Squibb Wohler, E.S., Hoover-Fong, J.E., Lisi, E., Stevens, E.L., Thomas, G.H., Leonard, J., Hamosh, A., Pevsner, J. (2010). "Genomic analysis of partial 21q monosomies with variable phenotypes." European Journal of Human Genetics **19(2)**: 235-238.

Robertson, E. (1987). "Embryo-derived stem cell lines. In Robertson, E. (ed.), Teratocarcinomas and embryonic stem cells – a practical approach." IRL Press, Oxford, UK: 77-112.

Roland, B., Cox, M., Hoar, D.I., Fowlowan, S.B., Robertson, A.S. (1990). "A familial interstitial deletion of the long arm of chromosome 21." Clinical Genetics **31**: 423-428.

Sago, H., Carlson, E.J., Smith, D.J., Kilbridge, J., Rubin, E.M.,. Mobley, W.C, Epstein C.J., Huang,T.T (1998). "Ts1Cje, a partial trisomy 16 mouse model for Down syndrome, exhibits learning and behavioral abnormalities." Protocols of National Academy of Science USA **95**: 6256-6261.

Sakata, K., Tamura, G., Nishizuka, S., Maesawa, C., Suzuki, Y., Iwaya, T., Terashima, M., Saito, K., Satodate, R. (1997). "Commonly deleted regions on the long arm of chromosome 21 in differentiated adenocarcinoma of the stomach." Genes, Chromosomes & Cancer **18**: 318-321.

Sato, M., Kawakami, T., Kondoh, M., Takiguchi, M., Kadota, Y., Himeno, S., Suzuki, S. (2010). "Development of high-fat-diet-induced obesity in female metallothionein-null mice." FASEB Journal **24**(7): 2375-2384.

Sato, S., Nakamura, Y., Tsuchiya, E. (1994). "Difference of allelotype between squamous cell carcinoma and adenocarcinoma of the lung." Cancer Research **54**: 5652-5655.

Sauer, B., Henderson, N. (1988). "Site-specific DNA recombination in mammalian cells by the Cre recombinase of bacteriophage P1." Proceedings of the National Academy of Sciences of the United States of America **85**(14): 5166-5170.

Saugier-Veber, P., Bonnet, C., Afenjar, A., Drouin-Garraud, V., Coubes, C., Fehrenbach, S., Holder-Espinasse, M., Roume, J., Malan, V., Portnoi, M.F., Jeanne, N., Baumann, C., Héron, D., David, A., Gérard, M., Bonneau, D., Lacombe, D., Cormier-Daire, V., Billette de Villemeur, T., Frébourg, T., Bürglen, L. (2007). "Heterogeneity of NSD1 alterations in 116 patients with Sotos syndrome." Human Mutation **28**(11): 1098-1107.

Shaffer, L.G., Lupski, J.R. (2009). "Molecular mechanisms for constitutional chromosomal rearrangements in humans " Annual Reviews in Genetics **34**: 297-329.

Skryabin, B.V., Gubar, L.V., Seeger, B., Pfeiffer, J., Handel, S., Robeck, T., Karpova, E., Rozhdestvensky, T.S., Brosius, J. (2007). "Deletion of the MBII-85 snoRNA gene cluster in mice results in postnatal growth retardation." PLoS Genetics **3**(12): 2529-2539.

Smith, A.C., McGavran, L., Robinson, J., Waldstein, G., Macfarlane, J., Zonona, J., Reiss, J., Lahr, M., Allen, L., Magenis, E. (1986). "Interstitial deletion of (17)(p11.2p11.2) in nine patients." American Journal of Medical Genetics **24**(3): 393-414.

Smith, A.G. (1991). "Culture and differentation of embryonic stem cells." Journal of Tissue Culture Methods **13**: 89-94.

Smyth, G.K. (2004). "Linear models and empirical bayes methods for assessing differential expression in microarray experiments." Statistical Applications in Genetics and Molecular Biology **3, No. 1, Article 3**: 1-26.

Sotos, J.F., Dodge, P.R., Muirhead, D., Crawford, J.D., Talbot, N.B. (1964). "Cerebral gigantism in childhood – a syndrome of excessively rapid growth with acromegalic features and a nonprogressive neurologic disorder." New England Journal of Medicine **271**: 109-116.

Stec, I., Nagl, S.B., van Ommen, G.J.B., den Dunnen, J.T. (2000). "The PWWP domain: a potential protein-protein interaction domain in nuclear proteins infuencing differentiation." FEBS Letters **473**: 1-5.

Stubbs, L., Carver, E.A., Cacheiro, N.L., Shelby, M., Generoso, W. (1997). "Generation and characterization of heritable reciprocal translocations in mice." Methods **13**(4): 397-408.

Sullivan, T., Escalante-Alcalde, D., Bhatt, H., Anver, M., Bhat, N., Nagashima, K., Stewart, C.L., Burke, B. (1999). "Loss of A-type lamin expression compromises nuclear envelope integrity leading to muscular dystrophy." Journal of Cell Biology **147**(5): 913-919.

Tartaglia, N.R., Howell, S., Sutherland, A., Wilson, R., Wilson, L. (2010). "A review of trisomy X (47,XXX)." Orphanet Journal of Rare Diseases **5**(1): 1-9.

Tatton-Brown, K., Douglas, J., Coleman, K., Baujat, G., Chandler, K., Clarke, A., Collins, A., Davies, S., Faravelli, F., Firth, H., Garrett, C., Hughes, H., Kerr, B., Liebelt, J., Reardon, W, Schaefer, G.B., Splitt, M., Temple, I.K., Waggoner, D., Weaver, D.D, Wilson, L., Cole, T., Cormier-Daire, V., Irrthum, A., Rahman, N. and on behalf of the Childhood Overgrowth Collaboration (2005). "Multiple mechanisms are implicated in the generation of 5q35 microdeletions in Sotos syndrome." Journal of Medical Genetics **42**(4): 307-313.

Tatton-Brown, K., Douglas, J., Coleman, K., Baujat, G., Cole, T.R.P., Das, S., Horn, D., Hughes, H.E., Temple, I.K., Faravelli, F., Waggoner, D., Tu¨rkmen, S., Cormier-Daire, V., Irrthum, A., Rahman, N. for the Childhood Overgrowth Collaboration (2005). "Genotype-phenotype associations in Sotos syndrome: an analysis of 266 individuals with NSD1 aberrations." American Journal of Human Genetics **77**: 193-204.

Tatton-Brown, K., Rahman, N. (2007). "Sotos syndrome." European Journal of Human Genetics **15**: 264-271.

Tinkel-Vernon, H., Finkemagel, S., Desposito, F., Pittore, C., Reynolds, K., Sciorra, L. (2003). "Patient with a deletion of chromosome 21q and minimal phenotype." American Journal of Medical Genetics **120A**(1): 142-143.

Tsai, T.F., Jiang, J.H., Bressler, J., Armstrong, D., Beaudet, A.L. (1999). "Paternal deletion fromSnrpn to Ube3a in the mouse causes hypotonia, growth retardation and partial lethality and provides evidence for a gene contributing to Prader–Willi syndrome." Human Molecular Genetics **8**(8): 1357-1364.

Tucker, M.E., Garringer, H.J., Weaver, D.D. (2007). "Phenotypic spectrum of mosaic trisomy 18: Two new patients, a literature review, and

counseling issues." American Journal of Medical Genetics Part A **143A**(5): 505-517.

Tunster, S.J., Van de Pette, M., John, R.M. (2011). "Fetal overgrowth in the Cdkn1c mouse model of Beckwith-Wiedemann syndrome." Disease Models & Mechanisms **July 4.**: 1-8.

Umans, L., Overbergh, L., Serneels, L., Tesseur, I., Van Leuven, F. (1999). "Analysis of expression of genes involved in apolipoprotein e-based lipoprotein metabolism in pregnant mice deficient in the receptor-associated protein, the low density lipoprotein receptor, or apolipoprotein E1." Biology of Reproduction **61**: 1216-1225.

Valero, R., Marfany, G., Gonzalez-Angulo, O., Gonzalez-Gonzalez, G., Puelles, L., Gonzalez-Duarte, R. (1999). "USP25, a novel gene encoding a deubiquitinating enzyme, is located in the gene-poor region 21q11.2." Genomics **62**: 395-405.

Van der Weyden, L., Shaw-Smith, C., Bradley, A (2009). "Chromosome engineering in ES cells." Methods in Molecular Biology **530**: 49-77.

Vani Rayasam, G., Wendling, O., Angrand, P.O., Mark, M., Niederreither, K., Song, L., Lerouge, T., Hager, G.L., Chambon, P., Losson, R. (2003). "NSD1 is essential for early post-implantation development and has a catalytically active SET domain." European Molecular Biology Organisation Journal **22**(12): 3153-3163.

Visser, R., Shimokawa, O., Harada, N., Kinoshita, A., Ohta, T., Niikawa, N., Matsumoto, N. (2005). "Identification of a 3.0-kb major recombination hotspot in patients with Sotos syndrome who carry a common 1.9-Mb microdeletion." American Journal of Human Genetics **76**: 52-67.

Wakui, K., Toyoda, A., Kubota, T., Hidaka, E., Ishikawa, M., Katsuyama, T., Sakaki, Y., Hattori, M., Fukushima Y. (2002). "Familial 14-Mb deletion at 21q11.2-q21.3 and variable phenotypic expression." Journal of Human Genetics **47(10)**: 511-516.

Walz, K., Caratini-Rivera, S., Bi, W., Fonseca, P., Mansouri, D.L., Lynch, J., Vogel, H., Noebels, J.L., Bradley, A., Lupski, J.R. (2003). "Modeling del(17)(p11.2p11.2) and dup(17)(p11.2p11.2) contiguous gene syndromes by chromosome engineering in mice: phenotypic

consequences of gene dosage imbalance." Molecular and Cellular Biology **23**(10): 3646-3655.

Walz, K., Spencer, C., Kaasik, K., Lee, C.C., Lupski, J.R., Paylor, R. (2003). "Behavioral characterization of mouse models for Smith-Magenis syndrome and dup(17)(p11.2p11.2)." Human Molecular Genetics **13**(4): 367-378.

Wang, D., Stewart, A.K, Zhuang, L., Zhu, Y., Wang, Y., Shi, C., Keating, A., Slutsky, A., Zhang, H., Wen, X.Y. (2010). "Enhanced adaptive immunity in mice lacking the immunoinhibitory adaptor Hacs1." FASEB Journal **24**(3): 947-956.

Waterston, R.H., et al. (2002). "Initial sequencing and comparative analysis of the mouse genome." Nature **402**: 520-562.

Wen, X.Y., Hegele, R.A., Wang, J., Wang, D.Y., Cheung, J., Wilson, M., Yahyapour, M., Bai, Y., Zhuang, L., Skaug, J., Young, T.K., Connelly, P.W., Koop, B.F.,Tsui, L.C., Stewart, A.K. (2003). "Identification of a novel lipase gene mutated in lpd mice with hypertriglyceridemia and associated with dyslipidemia in humans." Human Molecular Genetics **12**(10).

Weyer, C., Wolford, J.K., Hanson, R.L., Foley, J.E., Tataranni, P.A., Bogardus, C., Pratley, R.E. (2001). "Subcutaneous abdominal adipocyte size, a predictor of type 2 diabetes, is linked to chromosome 1q21– q23 and is associated with a common polymorphism in LMNA in Pima Indians." Molecular Genetics and Metabolism **72**: 231-238.

White, R., Leonardsson, G., Rosewell, I., Jacobs, M.A., Milligan, S., Parker, M. (2000). "The nuclear receptor co-repressor Nrip1 (RIP140) is essential for female fertility." Nature Medicine **6**(12): 1368-1374.

Wikström, A.M., Dunkel, L. (2011). "Klinefelter syndrome." Best Practice & Research Clinical Endocrinology & Metabolism **25**(2): 239-250.

Williams, J.C.P., Barratt-Boyes, B.G., Lowe, J.B. (1961). "Supravalvular aortic stenosis." Circulation **24**: 1311-1318.

Willnow, T.E., Armstrong, S.A., Hammer, R.E., Herz, J. (1995). "Functional expression of low density lipoprotein receptor-related protein is controlled by receptor-associated protein in vivo." Proceedings of the National Academy of Sciences **92**: 4537-4541.

Wojtanik, K.M., Edgemon, K., Viswanadha, S., Lindsey, B., Haluzik, M., Chen, W., Poy, G., Reitman, M., Londos, C. (2009). "The role of LMNA in adipose: a novel mouse model of lipodystrophy based on the Dunnigan-type familial partial lipodystrophy mutation." The Journal of Lipid Research **50**(6): 1068-1079.

Yabuuchi, H., Takayanagi, S., Yoshinaga, K., Taniguchi, N., Aburatani, H., Ishikawa, T. (2002). "ABCC13, an unusual truncated ABC transporter, is highly expressed in fetal human liver." Biochemical and Biophysical Research Communications **299**: 410-417.

Yamada, H., Yanagisawa, K., Tokumaru, S., Taguchi, A., Nimura, Y., Osada, H., Nagino, M., Takahashi, T. (2008). "Detailed characterization of a homozygously deleted region corresponding to a candidate tumor suppressor locus at 21q11-21 in human lung cancer." Genes, Chromosomes & Cancer **47**: 810-818.

Yamagata, N., Furuno, K., Sonoda, M., Sugimura, H., Yamamoto, K. (2008). "Stomach cancer-derived del223V-226L mutation of the STCH gene causes loss of sensitization to TRAIL-mediated apoptosis." Biochemical and Biophysical Research Communications **376**(3): 499-503.

Yamamoto, N., Mizoe, J., Numasawa, H., Tsujii, H., Shibahara, T., Noma, H. (2003). "Allelic loss on chromosomes 2q, 3p and 21q: possibly a poor prognostic factor in oral squamous cell carcinoma." Oral Oncology **39**(8): 796-805.

Yan, J., Keener, V.W, Bi, W., Walz, K., Bradley, A., Justice, M.J., Lupski, J.R. (2004). "Reduced penetrance of craniofacial anomalies as a function of deletion size and genetic background in a chromosome engineered partial mouse model for Smith-Magenis syndrome." Human Molecular Genetics **13**(21): 2613-2624.

Yang, S.H., Bergo, M.O., Toth, J.I., Qiao, X., Hu, Y., Sandoval, S., Meta, M., Bendale, P., Gelb, M.H., Young, S.G., Fong, L.G. (2005). "Blocking protein farnesyltransferase improves nuclear blebbing in mouse fibroblasts with a targeted Hutchinson–Gilford progeria syndrome mutation." Proceedings of the National Academy of Sciences **102** (29): 10291–10296.

Yang, T., Adamson, T.E., Resnick, J.L., Leff, S., Wevrick, R., Francke, U., Jenkins, N.A., Copeland, N.G., Brannan, C.I. (1998). "A mouse model for Prader-Willi syndrome imprinting-centre mutations." Nature Genetics **19**(1): 25-31.

Yang, Y.H., Dudoit, S., Luu, P., Lin, D.M., Peng, V., Ngai, J., Speed, T.P. (2001). "Normalization ofr cDNA microarray data: a robust composite method addressing single and multiple slide systematic variation." Nucleic Acids Research **30, No. 4, e15**: 1-10.

Yokoyama, T., Silversides, D.W., Waymire, K.G., Kwon, B.S., Takeuchi, T., Overbeek, P.A. (1990). "Conserved cysteine to serine mutation in tyrosinase is responsible for the classical albino mutation in laboratory mice." Nucleic Acids Research **18**: 7293-7298.

Young, E.J., Lipina, T., Tam, E., Mandel, A., Clapcote, S.J., Bechard, A.R., Chambers, J., Mount, H.T.J., Fletcher, P.J., Roder, J.C., Osborne, L.R. (2008). "Reduced fear and aggression and altered serotonin metabolism in Gtf2ird1-targeted mice." Genes, Brain and Behavior **7**(2): 224-234.

Yu, T., Clapcote, S.J., Li, Z., Liu, C., Pao, A., Bechard, A.R., Carattini-Rivera, S., Matsui, S.I., Roder, J.C., Baldini, A., Mobley, W.C., Bradley, A., Yu, E.Y. (2010). "Deficiencies in the region syntenic to human 21q22.3 cause cognitive deficits in mice." Mammalian Genome **21(5-6)**: 258-267.

Zhao, L.J., Jiang, H., Papasian, C.J., Maulik, D., Drees,B., Hamilton, J., Deng, H.W. (2008). "Correlation of obesity and osteoporosis: effect of fat mass on the determination of osteoporosis." Journal of bone and mineral research **23**(1): 17-29.

Zheng, B., Mills, A.A., Bradley, A. (1999). "A system for rapid generation of coat color-tagged knockouts and defined chromosomal rearrangements in mice." Nucleic Acids Research **27**(11): 2354-2360.

Zheng, B., Sage, M., Cai, W.W., Thompson, D.M., Tavsanli, B.C., Cheah, Y.C., Bradley, A. (1999). "Engineering a mouse balancer chromosome." Nature Genetics **22**: 375-378.

Zheng, B., Sage, M., Sheppeard, E.A., Jurecic, V., Bradley, A. (2000). "Engineering mouse chromosomes with Cre-loxP: range, efficiency, and somatic applications." Molecular Cell Biology **20**: 648-655.