6. Literaturverzeichnis

Baujat, G., Rio, M., Rossignol, S., Sanlaville, D., Lyonnet, S., Le Merrer, M., Munnich, A., Gicquel, C., Colleaux, L. and Cormier-Daire, V. (2005). "Clinical and molecular overlap in overgrowth syndromes." Am J Med Genet C Semin Med Genet 137(1): 4-11.

Demirhan O*, Turkmen S*, Schwabe GC, Soyupak S, Akgul E, Tastemir D, Karahan D, Mundlos S, Lehmann K. (2005) A homozygous BMPR1B mutation causes a new subtype of acromesomelic chondrodysplasia with genital anomalies.J Med Genet Apr;42(4):314-7.

Douglas, J., Hanks, S., Temple, I. K., Davies, S., Murray, A., Upadhyaya, M., Tomkins, S., Hughes, H. E., Cole, T. R. and Rahman, N. (2003). "NSD1 mutations are the major cause of Sotos syndrome and occur in some cases of Weaver syndrome but are rare in other overgrowth phenotypes." Am J Hum Genet 72(1): 132-43.

Erasmus CE, Beems T, Rotteveel JJ. (2004) Frontal ataxia in childhood. Neuropediatrics 35 (6) :368–70

Everman DB, Bartels CF, Yang Y, Yanamandra N, Goodman FR, Mendoza-Londono JR, Savarirayan R, White SM, Graham JM Jr, Gale RP, Svarch E, Newman WG, Kleckers AR, Francomano CA, Govindaiah V, Singh L, Morrison S, Thomas JT, Warman ML.(2002) The mutational spectrum of brachydactyly type C. Am J Med Genet 112 (3) :291–6

Faiyaz-Ul-Haque M, Ahmad W, Zaidi SH, Haque S, Teebi AS, Ahmad M, Cohn DH, Tsui LC. (2002) Mutation in the cartilage-derived morphogenetic protein-1 (CDMP1) gene in a kindred affected with fibular hypoplasia and complex brachydactyly (DuPan syndrome). Clin Genet 61 (6) :454–8.

Fukuyama H, Ouchi Y, Matsuzaki S, Nagahama Y, Yamauchi H, Ogawa M, Kimura J, Shibasaki H. (1997) Brain functional activity during gait in normal subjects: a SPECT study. Neurosci Lett 228 (3) :183–6

Klein C.(2005) Movement disorders: classifications. J Inherit Metab Dis 28 (3) :425–39

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 Ha, H. A., Kinoshita, A., Mizuguchi, T., Yoshiura Ki, K., Ohta, T., Kishino, T., Fukushima, Y., Niikawa, N. and Matsumoto, N.. (2002) "Haploinsufficiency of NSD1 causes Sotos syndrome." Nat Genet **30**(4): 365-6.

Mulsant P, Lecerf F, Fabre S, Schibler L, Monget P, Lanneluc I, Pisselet C, Riquet J, Monniaux D, Callebaut I, Cribiu E, Thimonier J, Teyssier J, Bodin L, Cognie Y, Chitour N, Elsen J-M. (2001)Mutation in bone morphogenetic protein receptor-IB is associated with increased ovulation rate in Booroola Merino ewes. Proc Natl Acad Sci U S A 98 (9) :5104–9

Polinkovsky A, Robin NH, Thomas JT, Irons M, Lynn A, Goodman FR, Reardon W, Kant SG, Brunner HG, van der Burgt I, Chitayat D, McGaughran J, Donnai D, Luyten FP, Warman ML. (1997)Mutations in CDMP1 cause autosomal dominant brachydactyly type C. Nat Genet 17 (1) :18–9

Saiki RK, Scharf S, Faloona F, Mullis KB, Horn GT, Erlich HA,Arnheim N. (1985) Enzymatic amplification of beta-globin genomic sequences and restriction site analysis for diagnosis of sickle cell anemia. Science Dec 20;230(4732):1350-4.

Sambrook, J., Fritsch, E. F. and Maniatis, T. (1989). "Molecular cloning-a laboratory manual." Cold Spring Harbor, New York.

Sanger, F., Nicklen, S. and Coulsen, A.R. (1977). "DNA sequencing with chain-terminating inhibitors." Proc Natl Acad Sci U S A 74(12): 5463-7.

Sotos JF, Dodge PR, Muirhead D, Crawford JD, Talbot NB (1964) Cerebral gigantism in childhood. N Engl J Med 271:109–116.

Sutherland DH, Olshen R, Cooper L, Woo SL. (1980)The development of mature gait. J Bone Joint Surg Am 62 (3) :336–53

ten Donkelaar HJ, Lammens M, Wesseling P, Thijssen HO, Renier WO. (2003) Development and developmental disorders of the human cerebellum. J Neurol 250(9):1025-36

Thach WT, Bastian AJ. (2004) Role of the cerebellum in the control and adaptation of gait in health and disease. Prog Brain Res;**143**:353–66

Thomas JT, Kilpatrick MW, Lin K, Erlacher L, Lembessis P, Costa T, Tsipouras P, Luyten FP.(1997) Disruption of human limb morphogenesis by a dominant negative mutation in CDMP1. Nat Genet 17 (1) :58–64

Thomas JT, Lin K, Nandedkar M, Camargo M, Cervenka J, Luyten FP. (1996) A human chondrodysplasia due to a mutation in a TGF-beta superfamily member. Nat Genet 12 (3) :315–7

Turkmen S, Gillessen-Kaesbach, G., Meinecke, P., Albrecht, B., Neumann, L. M., Hesse, V.,

Palanduz, S., Balg, S., Majewski, F., Fuchs, S., Zschieschang, P., Greiwe, M., Mennicke, K., Kreuz, F. R., Dehmel, H. J., Rodeck, B., Kunze, J., Tinschert, S., Mundlos, S. and Horn, D. (2003). "Mutations in NSD1 are responsible for Sotos syndrome, but are not a frequent finding in other overgrowth phenotypes." Eur J Hum Genet 11(11): 858-65.

Turkmen S, Demirhan O, Hoffmann K, Diers A, Zimmer C, Sperling K, Mundlos S. (2006) Cerebellar hypoplasia and quadrupedal locomotion in humans as a recessive trait mapping to chromosome 17p. J Med Genet May;43(5):461-4.

Wilson T, Wu XY, Juengel JL, Ross IK, Lumsden JM, Lord EA, Dodds KG, Walling GA, McEwan JC, O'Connell AR, McNatty KP, Montgomery GW. (2001) Highly prolific Booroola sheep have a mutation in the intracellular kinase domain of bone morphogenetic protein IB receptor (ALK-6) that is expressed in both oocytes and granulosa cells. Biol Reprod 64 (4) :1225–35

Yi SE, LaPolt PS, Yoon BS, Chen JY-C, Lu JKH, Lyons KM. (2001) The type I BMP receptor BmprIB is essential for female reproductive function. Proc Natl Acad Sci U S A **98** (14) :7994–9