

## 6. Literaturverzeichnis

- Aoki, Y., Niihori, T., Kawame, H., Kurosawa, K., Ohashi, H., Tanaka, Y., Filocamo, M., Kato, K., Suzuki, Y., Kure, S. and Matsubara, Y. (2005). "Germline mutations in HRAS proto-oncogene cause Costello syndrome." *Nat Genet* **37**(10): 1038-40.
- Bartsch, O., Hinkel, G. K., Petersen, M. B., Konig, U., Bugge, M., Mikkelsen, M., Avramopoulos, D., Morris, M. and Antonarakis, S. E. (1997). "A large family with subtelomeric translocation t(18;21)(q23;q22.1) and molecular breakpoint in the Down syndrome critical region." *Hum Genet* **100**(5-6): 669-75.
- 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.
- Belloni, E., Martucciello, G., Verderio, D., Ponti, E., Seri, M., Jasonni, V., Torre, M., Ferrari, M., Tsui, L. C. and Scherer, S. W. (2000). "Involvement of the HLXB9 homeobox gene in Curarino syndrome." *Am J Hum Genet* **66**(1): 312-9.
- Benzacken, B., Siffroi, J. P., Le Bourhis, C., Krabchi, K., Joye, N., Maschino, F., Viguie, F., Soulie, J., Gonzales, M., Migne, G., Bucourt, M., Encha-Razavi, F., Carbillon, L. and Taillemite, J. L. (1997). "Different proximal and distal rearrangements of chromosome 7q associated with holoprosencephaly." *J Med Genet* **34**(11): 899-903.
- Cohen, M. M., Jr., Hall, B. D., Smith, D. W., Graham, C. B. and Lampert, K. J. (1973). "A new syndrome with hypotonia, obesity, mental deficiency, and facial, oral, ocular, and limb anomalies." *J Pediatr* **83**(2): 280-4.
- Curry, C. J., Stevenson, R. E., Aughton, D., Byrne, J., Carey, J. C., Cassidy, S., Cunniff, C., Graham, J. M., Jr., Jones, M. C., Kaback, M. M., Moeschler, J., Schaefer, G. B., Schwartz, S., Tarleton, J. and Opitz, J. (1997). "Evaluation of mental retardation: recommendations of a Consensus Conference: American College of Medical Genetics." *Am J Med Genet* **72**(4): 468-77.
- de Vries, B. B., Pfundt, R., Leisink, M., Koolen, D. A., Vissers, L. E., Janssen, I. M., Reijmersdal, S., Nillesen, W. M., Huys, E. H., Leeuw, N., Smeets, D., Sistermans, E. A., Feuth, T., van Ravenswaaij-Arts, C. M., van Kessel, A. G., Schoenmakers, E. F., Brunner, H. G. and Veltman, J. A. (2005). "Diagnostic genome profiling in mental retardation." *Am J Hum Genet* **77**(4): 606-16.
- Delabar, J. M., Theophile, D., Rahmani, Z., Chettouh, Z., Blouin, J. L., Prieur, M., Noel, B. and Sinet, P. M. (1993). "Molecular mapping of twenty-four features of Down syndrome on chromosome 21." *Eur J Hum Genet* **1**(2): 114-24.
- 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.
- Frints, S. G., Schoenmakers, E. F., Smeets, E., Petit, P. and Fryns, J. P. (1998). "De novo 7q36 deletion: breakpoint analysis and types of holoprosencephaly." *Am J Med Genet* **75**(2): 153-8.
- Fryns, J. P., Legius, E., Devriendt, K., Meire, F., Standaert, L., Baten, E. and Van den Berghe, H. (1996). "Cohen syndrome: the clinical symptoms and stigmata at a young age." *Clin Genet* **49**(5): 237-41.

- Garcia-Heras, J. and Rao, P. N. (1999). "A brief review of cryptic duplications of 21q as an emerging cause of Down syndrome: practical considerations for accurate detection." *Clin Genet* **55**(3): 207-11.
- Gripp, K. W., Lin, A. E., Stabley, D. L., Nicholson, L., Scott, C. I., Jr., Doyle, D., Aoki, Y., Matsubara, Y., Zackai, E. H., Lapunzina, P., Gonzalez-Meneses, A., Holbrook, J., Agresta, C. A., Gonzalez, I. L. and Sol-Church, K. (2006). "HRAS mutation analysis in Costello syndrome: genotype and phenotype correlation." *Am J Med Genet A* **140**(1): 1-7.
- Happle, R. (1993). "Klippel-Trenaunay syndrome: is it a paradigmatic trait?" *Br J Dermatol* **128**(4): 465-6.
- Happle, R. (1993). "Mosaicism in human skin. Understanding the patterns and mechanisms." *Arch Dermatol* **129**(11): 1460-70.
- Hennies, H. C., Rauch, A., Seifert, W., Schumi, C., Moser, E., Al-Taji, E., Tariverdian, G., Chrzanowska, K. H., Krajewska-Walasek, M., Rajab, A., Giugliani, R., Neumann, T. E., Eckl, K. M., Karbasiyan, M., Reis, A. and Horn, D. (2004). "Allelic heterogeneity in the COH1 gene explains clinical variability in Cohen syndrome." *Am J Hum Genet* **75**(1): 138-45.
- Horn, D., Chyrek, M., Kleier, S., Luttenberg, S., Bolz, H., Hinkel, G. K., Korenke, G. C., Riess, A., Schell-Apacik, C., Tinschert, S., Wieczorek, D., Gillessen-Kaesbach, G. and Kutsche, K. (2005). "Novel mutations in BCOR in three patients with oculo-facio-cardio-dental syndrome, but none in Lenz microphthalmia syndrome." *Eur J Hum Genet* **13**(5): 563-9.
- Horn, D., Happle, R., Neitzel, H. and Kunze, J. (2002). "Pigmentary mosaicism of the hyperpigmented type in two half-brothers." *Am J Med Genet* **112**(1): 65-9.
- Horn, D., Krebssova, A., Kunze, J. and Reis, A. (2000). "Homozygosity mapping in a family with microcephaly, mental retardation, and short stature to a Cohen syndrome region on 8q21.3-8q22.1: redefining a clinical entity." *Am J Med Genet* **92**(4): 285-92.
- Horn, D., Majewski, F., Hildebrandt, B. and Korner, H. (1995). "Pallister-Killian syndrome: normal karyotype in prenatal chorionic villi, in postnatal lymphocytes, and in slowly growing epidermal cells, but mosaic tetrasomy 12p in skin fibroblasts." *J Med Genet* **32**(1): 68-71.
- Horn, D., Neitzel, H., Tonnes, H., Kalscheuer, V., Kunze, J., Hinkel, G. K. and Bartsch, O. (2003). "Familial MCA/MR syndrome due to inherited submicroscopic translocation t(18;21)(q22.1q21.3) with breakpoint at the Down syndrome critical region." *Am J Med Genet A* **117**(3): 236-44.
- Horn, D., Rommeck, M., Sommer, D. and Korner, H. (1997). "Phylloid pigmentary pattern with mosaic trisomy 13." *Pediatr Dermatol* **14**(4): 278-80.
- Horn, D., Tonnes, H., Neitzel, H., Wahl, D., Hinkel, G. K., von Moers, A. and Bartsch, O. (2004). "Minimal clinical expression of the holoprosencephaly spectrum and of Curranino syndrome due to different cytogenetic rearrangements deleting the Sonic Hedgehog gene and the HLXB9 gene at 7q36.3." *Am J Med Genet A* **128**(1): 85-92.
- Horn, D., Weschke, B., Zweier, C. and Rauch, A. (2004). "Facial phenotype allows diagnosis of Mowat-Wilson syndrome in the absence of Hirschsprung disease." *Am J Med Genet A* **124**(1): 102-4.
- Horneff, G., Majewski, F., Hildebrand, B., Voit, T. and Lenard, H. G. (1993). "Pallister-Killian syndrome in older children and adolescents." *Pediatr Neurol* **9**(4): 312-5.
- Kivitie-Kallio, S. and Norio, R. (2001). "Cohen syndrome: essential features, natural history, and heterogeneity." *Am J Med Genet* **102**(2): 125-35.

- Kivitie-Kallio, S., Rajantie, J., Juvonen, E. and Norio, R. (1997). "Granulocytopenia in Cohen syndrome." *Br J Haematol* **98**(2): 308-11.
- Knight, L. A., Yong, M. H., Tan, M. and Ng, I. S. (1996). "Subtle translocation (18;21) confirmed by FISH in a patient with Down syndrome." *Clin Genet* **50**(5): 430-2.
- Knight, S. J. and Flint, J. (2000). "Perfect endings: a review of subtelomeric probes and their use in clinical diagnosis." *J Med Genet* **37**(6): 401-9.
- Knight, S. J., Regan, R., Nicod, A., Horsley, S. W., Kearney, L., Homfray, T., Winter, R. M., Bolton, P. and Flint, J. (1999). "Subtle chromosomal rearrangements in children with unexplained mental retardation." *Lancet* **354**(9191): 1676-81.
- Kochling, J., Karbasiyan, M. and Reis, A. (2001). "Spectrum of mutations and genotype-phenotype analysis in Currarino syndrome." *Eur J Hum Genet* **9**(8): 599-605.
- Kolehmainen, J., Black, G. C., Saarinen, A., Chandler, K., Clayton-Smith, J., Traskelin, A. L., Perveen, R., Kivitie-Kallio, S., Norio, R., Warburg, M., Fryns, J. P., de la Chapelle, A. and Lehesjoki, A. E. (2003). "Cohen syndrome is caused by mutations in a novel gene, COH1, encoding a transmembrane protein with a presumed role in vesicle-mediated sorting and intracellular protein transport." *Am J Hum Genet* **72**(6): 1359-69.
- Kolehmainen, J., Norio, R., Kivitie-Kallio, S., Tahvanainen, E., de la Chapelle, A. and Lehesjoki, A. E. (1997). "Refined mapping of the Cohen syndrome gene by linkage disequilibrium." *Eur J Hum Genet* **5**(4): 206-13.
- Kratz, C. P., Schubbert, S., Bollag, G., Niemeyer, C. M., Shannon, K. M. and Zenker, M. (2006). "Germline mutations in components of the ras signaling pathway in noonan syndrome and related disorders." *Cell Cycle* **5**(15): 1607-11.
- 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.
- Lynch, S. A., Bond, P. M., Copp, A. J., Kirwan, W. O., Nour, S., Balling, R., Mariman, E., Burn, J. and Strachan, T. (1995). "A gene for autosomal dominant sacral agenesis maps to the holoprosencephaly region at 7q36." *Nat Genet* **11**(1): 93-5.
- Lynch, S. A., Wang, Y., Strachan, T., Burn, J. and Lindsay, S. (2000). "Autosomal dominant sacral agenesis: Currarino syndrome." *J Med Genet* **37**(8): 561-6.
- Masuno, M., Fukushima, Y., Sugio, Y., Ikeda, M. and Kuroki, Y. (1990). "Two unrelated cases of single maxillary central incisor with 7q terminal deletion." *Jinrui Idengaku Zasshi* **35**(4): 311-7.
- Mendez, H. M., Paskulin, G. A. and Vallandro, C. (1985). "The syndrome of retinal pigmentary degeneration, microcephaly, and severe mental retardation (Mirhosseini-Holmes-Walton syndrome): report of two patients." *Am J Med Genet* **22**(2): 223-8.
- Mirhosseini, S. A., Holmes, L. B. and Walton, D. S. (1972). "Syndrome of pigmentary retinal degeneration, cataract, microcephaly, and severe mental retardation." *J Med Genet* **9**(2): 193-6.
- Mochida, G. H., Rajab, A., Eyaid, W., Lu, A., Al-Nouri, D., Kosaki, K., Noruzinia, M., Sarda, P., Ishihara, J., Bodell, A., Apse, K. and Walsh, C. A. (2004). "Broader geographical spectrum of Cohen syndrome due to COH1 mutations." *J Med Genet* **41**(6): e87.
- Moeschler, J. B. and Shevell, M. (2006). "Clinical genetic evaluation of the child with mental retardation or developmental delays." *Pediatrics* **117**(6): 2304-16.

- Mowat, D. R., Wilson, M. J. and Goossens, M. (2003). "Mowat-Wilson syndrome." *J Med Genet* **40**(5): 305-10.
- Ng, D., Thakker, N., Corcoran, C. M., Donnai, D., Perveen, R., Schneider, A., Hadley, D. W., Tifft, C., Zhang, L., Wilkie, A. O., van der Smagt, J. J., Gorlin, R. J., Burgess, S. M., Bardwell, V. J., Black, G. C. and Biesecker, L. G. (2004). "Oculofaciocardiodental and Lenz microphthalmia syndromes result from distinct classes of mutations in BCOR." *Nat Genet* **36**(4): 411-6.
- Niihori, T., Aoki, Y., Narumi, Y., Neri, G., Cave, H., Verloes, A., Okamoto, N., Hennekam, R. C., Gillessen-Kaesbach, G., Wieczorek, D., Kavamura, M. I., Kurosawa, K., Ohashi, H., Wilson, L., Heron, D., Bonneau, D., Corona, G., Kaname, T., Naritomi, K., Baumann, C., Matsumoto, N., Kato, K., Kure, S. and Matsubara, Y. (2006). "Germline KRAS and BRAF mutations in cardio-facio-cutaneous syndrome." *Nat Genet* **38**(3): 294-6.
- Norio, R., Raitta, C. and Lindahl, E. (1984). "Further delineation of the Cohen syndrome; report on chorioretinal dystrophy, leukopenia and consanguinity." *Clin Genet* **25**(1): 1-14.
- Opitz, C., Horn, D., Lehmann, R., Dimitrova, T. and Fasmers-Henke, K. (1998). "Oculo-facio-cardio-dental (OFCD) syndrome." *J Orofac Orthop* **59**(3): 178-85.
- Opitz, J.M., Kaveggia, E.G., Laxova, R., Pallister, P.D. (1982). "The diagnosis and prevention of severe mental retardation." Vol II. Proceedings, International Conference on Preventable Aspects of Genetic Morbidity. Cairo, pp 117-38.
- Opitz, J. M., Weaver, D. W. and Reynolds, J. F., Jr. (1998). "The syndromes of Sotos and Weaver: reports and review." *Am J Med Genet* **79**(4): 294-304.
- Rauch, A., Hoyer, J., Guth, S., Zweier, C., Kraus, C., Becker, C., Zenker, M., Huffmeier, U., Thiel, C., Ruschendorf, F., Nurnberg, P., Reis, A. and Trautmann, U. (2006). "Diagnostic yield of various genetic approaches in patients with unexplained developmental delay or mental retardation." *Am J Med Genet A* **140**(19): 2063-74.
- Roessler, E., Belloni, E., Gaudenz, K., Jay, P., Berta, P., Scherer, S. W., Tsui, L. C. and Muenke, M. (1996). "Mutations in the human Sonic Hedgehog gene cause holoprosencephaly." *Nat Genet* **14**(3): 357-60.
- Schinzel, A. (1991). "Tetrasomy 12p (Pallister-Killian syndrome)." *J Med Genet* **28**(2): 122-5.
- Schubbert, S., Zenker, M., Rowe, S. L., Boll, S., Klein, C., Bollag, G., van der Burgt, I., Musante, L., Kalscheuer, V., Wehner, L. E., Nguyen, H., West, B., Zhang, K. Y., Sistermans, E., Rauch, A., Niemeyer, C. M., Shannon, K. and Kratz, C. P. (2006). "Germline KRAS mutations cause Noonan syndrome." *Nat Genet* **38**(3): 331-6.
- Schulze, B. R., Horn, D., Kobelt, A., Tariverdian, G. and Stellzig, A. (1999). "Rare dental abnormalities seen in oculo-facio-cardio-dental (OFCD) syndrome: three new cases and review of nine patients." *Am J Med Genet* **82**(5): 429-35.
- Seifert, W., Holder-Espinasse, M., Spranger, S., Hoeltzenbein, M., Rossier, E., Dollfus, H., Lacombe, D., Verloes, A., Chrzanowska, K. H., Maegawa, G. H., Chitayat, D., Kotzot, D., Huhle, D., Meinecke, P., Albrecht, B., Mathijssen, I., Leheup, B., Raile, K., Hennies, H. C. and Horn, D. (2006). "Mutational spectrum of COH1 and clinical heterogeneity in Cohen syndrome." *J Med Genet* **43**(5): e22.
- Shibuya, K., Kudoh, J., Minoshima, S., Kawasaki, K., Asakawa, S. and Shimizu, N. (2000). "Isolation of two novel genes, DSCR5 and DSCR6, from Down syndrome critical region on human chromosome 21q22.2." *Biochem Biophys Res Commun* **271**(3): 693-8.

- Tahvanainen, E., Norio, R., Karila, E., Ranta, S., Weissenbach, J., Sistonen, P. and de la Chapelle, A. (1994). "Cohen syndrome gene assigned to the long arm of chromosome 8 by linkage analysis." *Nat Genet* **7**(2): 201-4.
- Tartaglia, M., Kalidas, K., Shaw, A., Song, X., Musat, D. L., van der Burgt, I., Brunner, H. G., Bertola, D. R., Crosby, A., Ion, A., Kucherlapati, R. S., Jeffery, S., Patton, M. A. and Gelb, B. D. (2002). "PTPN11 mutations in Noonan syndrome: molecular spectrum, genotype-phenotype correlation, and phenotypic heterogeneity." *Am J Hum Genet* **70**(6): 1555-63.
- Tartaglia, M., Mehler, E. L., Goldberg, R., Zampino, G., Brunner, H. G., Kremer, H., van der Burgt, I., Crosby, A. H., Ion, A., Jeffery, S., Kalidas, K., Patton, M. A., Kucherlapati, R. S. and Gelb, B. D. (2001). "Mutations in PTPN11, encoding the protein tyrosine phosphatase SHP-2, cause Noonan syndrome." *Nat Genet* **29**(4): 465-8.
- Tatton-Brown, K., Douglas, J., Coleman, K., Baujat, G., Cole, T. R., Das, S., Horn, D., Hughes, H. E., Temple, I. K., Faravelli, F., Waggoner, D., Turkmen, S., Cormier-Daire, V., Irrthum, A. and Rahman, N. (2005). "Genotype-phenotype associations in Sotos syndrome: an analysis of 266 individuals with NSD1 aberrations." *Am J Hum Genet* **77**(2): 193-204.
- Tejada, M. I., Uribarren, A., Briones, P. and Vilaseca, M. A. (1992). "A further prenatal diagnosis of mosaic tetrasomy 12p (Pallister-Killian syndrome)." *Prenat Diagn* **12**(6): 529-34.
- 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.
- Wolf, G. (2006). "Qualitätssicherung Genetische Beratung. Ergebnisse der ersten Umfrage." *Med. Gen.* **18**: 229-34.
- Zenker, M., Lehmann, K., Schulz, A. L., Barth, H., Hansmann, D., Koenig, R., Korinthenberg, R., Kreiss-Nachtsheim, M., Meinecke, P., Morlot, S., Mundlos, S., Quante, A. S., Raskin, S., Schnabel, D., Wehner, L. E., Kratz, C. P., Horn, D. and Kutsche, K. (2007). "Expansion of the genotypic and phenotypic spectrum in patients with KRAS germline mutations." *J Med Genet* **44**(2): 131-5.
- Zweier, C., Albrecht, B., Mitulla, B., Behrens, R., Beese, M., Gillessen-Kaesbach, G., Rott, H. D. and Rauch, A. (2002). ""Mowat-Wilson" syndrome with and without Hirschsprung disease is a distinct, recognizable multiple congenital anomalies-mental retardation syndrome caused by mutations in the zinc finger homeo box 1B gene." *Am J Med Genet* **108**(3): 177-81.
- Zweier, C., Horn, D., Kraus, C. and Rauch, A. (2006). "Atypical ZFHX1B mutation associated with a mild Mowat-Wilson syndrome phenotype." *Am J Med Genet A* **140**(8): 869-72.