

5. Verzeichnis der eingebundenen Literatur

- Horn D, Krebsová A, Kunze J, Reis A. (2000). Homozygosity mapping in a family with microcephaly, mental retardation, and short stature to a Cohen syndrome region on chromosome 8q21.3-8q22.1: redefining a clinical entity. Am J Med Genet 92: 285-92. 6
- Hennies HC, Rauch A, Seifert W, Schumi C, Moser E, Al Taji E, Tariverdian G, Chrzanowska KH, Krajewska-Walasek M, Rajab A, Giugliani R, Neumann TE, Eckl KM, Karbasiyan M, Reis A, **Horn D**. (2004). Allelic heterogeneity in the COH1 gene explains clinical variability in Cohen syndrome. Am J Hum Genet 75: 138-45. 17
- Seifert W, Holder-Espinasse M, Spranger S, Hoeltzenbein M, Rossier E, Dollfus H, Lacombe D, Verloes A, Chrzanowska KH, Maegawa GHB, Chitayat D, Kotzot D, Huhle D, Meinecke P, Albrecht B, Mathijssen I, Leheup B, Raile K, Hennies HC, **Horn D**. (2006). Mutational spectrum of *COH1* and clinical heterogeneity in Cohen syndrome. J Med Genet 43: e22 25
- Zenker M, Lehmann K, Schulz AL, Barth H, Hansmann D, Koenig R, Korinthenberg R, Kreiß-Nachtsheim M, Meinecke P, Morlot S, Mundlos S, Quante AS, Raskin S, Schnabel D, Wehner LA, Kratz CP, **Horn D**^{*}, Kutsche K^{*}. (2007). Expansion of the genotypic and phenotypic spectrum in patients with KRAS germline mutations. J Med Genet. 44(2): 131-5.
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- Turkmen S, Gillessen-Kaesbach G, Meinecke P, Albrecht B, Neumann LM, Hesse V, Palanduz S, Balg S, Majewski F, Fuchs S, Zschieschang P, Greiwe M, Mennicke K, Kreuz F, Dehmel HJ, Kunze J, Tinschert S, Mundlos S, **Horn D**. (2003). NSD1 mutations are responsible for Sotos syndrome, but are not present in other overgrowth phenotypes. Eur J Hum Genet 11(11): 858-65. 40
- Tatton-Brown K, Douglas J, Coleman K, Baujat G, Cole TR, Das S, **Horn D**, Hughes HE, Temple IK, Faravelli F, Waggoner D, Turkmen S, Cormier-Daire V, Irrthum A, 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. 48
- Schulze BRB, **Horn D**, Kobelt A, Tariverdian G, 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: 429-35. 62
- **Horn D**, Chyrek M, Kleier S, Lüttgen S, Bolz H, Hinkel G, Korenke GC, Rieß A, Schell-Apacik C, Tinschert S, Wieczorek D, Gillessen-Kaesbach G, 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. 69

- **Horn D**, Weschke B, Zweier C, 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. 78
- Zweier C, **Horn D**, Kraus C, Rauch A: (2006). Atypical ZFHX1B mutation associated with a mild Mowat-Wilson syndrome phenotype. Am J Med Genet A 140(8): 869-72 81
- **Horn D**, Tönnies H, Neitzel H, Wahl D, Hinkel KG, von Moers A, 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. Am J Med Genet A. 128(1): 85-92. 87
- **Horn D**, Neitzel H, Tönnies H, Kalscheuer V, Kunze J, Hinkel GK, Bartsch O. (2003). Familial MCA/MR syndrome due to inherited submicroscopic translocation t(18;21)(q22.2q21.3) with breakpoint at the Down syndrome critical region. Am J Med Genet 117(3): 236-44. 97
- **Horn D**, Majewski F, Hildebrandt B, Körner 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: 68-71. 109
- **Horn D**, Rommeck M, Sommer D, Körner H. (1997). Phylloid pigmentary pattern with mosaic trisomy 13. Pediatr Dermatol 14 (4): 278-80. 113
- **Horn D**, Happle R, Neitzel H, Kunze J. (2002). Pigmentary mosaicism of the hyperpigmented type in two half-brothers. Am J Med Genet 112: 65-9. 116