Supplementary data

Appendix 1. HPO data source and HPO codes for all phenotypes related to preaxial polydactyly of the foot

Data source: http://compbio.charite.de/jenkins/job/hpo.annotations.monthly/
Dataset: ALL_SOURCES_ALL_FREQUENCIES_diseases_to_genes_to_phenotypes.txt
Last accession date: February 22, 2017

Search terms HPO database	HPO code
Preaxial polydactyly	HP:0100258
Preaxial foot polydactyly	HP:0001841
Foot polydactyly	HP:0001829
Polysyndactyly of hallux	HP:0005873
Duplication of the phalanx of the hallux	HP:0010066
Duplication of the proximal phalanx of the hallux	HP:0010093
Partial duplication of the distal phalanx of the hallux	HP:0010097
Broad hallux	HP:0010055
Broad phalanx of the toes	HP:0010174
Broad hallux phalanx	HP:0010059
Broad distal hallux	HP:0008111
Broad distal phalanx of the hallux	HP:0010077
Mirror image polydactyly	HP:0010689

Appendix 2. Genes related to the selected diseases

Disease	OMIM/Orphanet ID	Related gene
Acrocallosal Syndrome	OMIM:200990	*KIF7*
Apert Syndrome	OMIM:101200	*FGFR2*
Baraitser-Winter Syndrome	OMIM:243310	*ACTB*
Carpenter Syndrome (incl. subtypes)	OMIM:201000, ORPHANET:65759	*MEGF8, RAB23*
Chromosome 1Q21.1 Deletion Syndrome, 1.35-Mb	OMIM:612474	*GJA5, GJA8*
Congenital Heart Defects, Hamartomas of Tongue, and Polysyndactyly; CHDHTP	OMIM:217085	WDPCP
Craniofrontonasal Syndrome	OMIM:304110	EFNB1
Curry Jones Syndrome	ORPHANET:1553	SMO
Desbuquois Dysplasia	OMIM:251450	CANT1
Greig Cephalopolysyndactyly Syndrome	OMIM:175700	GLI3
Joubert Syndrome (incl. subtypes)	ORPHANET:475, ORPHANET:220493, ORPHANET:2318	*AHI1, ARL13B, B9D1,C5ORF42, CC2D2A, CEP104, CEP290, CEP41, CSPP1, HYLS1, INPP5E, KIAA0556, KIAA0586, MKS1, TCTN1, TCTN2, TMEM138, TMEM216, TMEM231, TMEM237, TMEM67, ZNF423*
Orofaciodigital Syndrome (incl. subtypes)	OMIM:277170, ORPHANET:2750, OMIM:258860	*C5ORF42, OFD1, TCTN3*
Pfeiffer Syndrome (incl. subtypes)	OMIM:101600, ORPHANET:93258, ORPHANET:93259, ORPHANET 93260	*FGFR1, FGFR2*
Polydactyly, Preaxial (incl. subtypes)	OMIM:174500	*LMBR1 (ZRS, Intron 5)*
Robinow-Sorauf Syndrome	OMIM:180750	*TWIST1*
Rubinstein-Taybi Syndrome (incl. subtypes)	OMIM:180849, OMIM:613684	*CREBBP, EP300*
Short-Rib Thoracic Dysplasia (incl. subtypes)	OMIM:263520	*NEK1*
Synpolydactyly (incl. subtypes)	OMIM:186000	*HOXD13*
Temple-Baraitser Syndrome	OMIM:611816	*KCNH1*