Panelinformation Kraniosynostos in-silico

Version: 2.0

I in-silicopanelanalysen ingår även MLPA-analys som detekterar förlust (deletioner) eller tillskott (duplikationer) av genetiskt material i TWIST1, FGFR1, FGFR2, FGFR3, RUNX2, MSX2, ALX1 (NM_006982.2), ALX3 (NM_006492.2), ALX4 och EFNB1.

Gen	Associerad fenotyp, MIM# (OMIM)	Nedärvning
ALX4	Frontonasal dysplasia 2, 613451	AR
	Parietal foramina 2, 609597	AD
	{Craniosynostosis 5, susceptibility to}, 615529	AD
ASXL1	Bohring-Opitz syndrome, 605039	AD
	Myelodysplastic syndrome, somatic, 614286	
BMP4	Microphthalmia, syndromic 6, 607932	AD
	Orofacial cleft 11, 600625	
CD96	C syndrome, 211750	AD
CDC45	Meier-Gorlin syndrome 7, 617063	AR
COLEC11	3MC syndrome 2, 265050	AR
CYP26B1	Craniosynostosis with radiohumeral fusions and other skeletal and craniofacial anomalies, 614416	
EFNB1	Craniofrontonasal dysplasia, 304110	XLD
ERF	Chitayat syndrome, 617180	AD
	Craniosynostosis 4, 600775	AD
FGFR1	Encephalocraniocutaneous lipomatosis, 613001	SMo
	Hartsfield syndrome, 615465	AD
	Hypogonadotropic hypogonadism 2 with or without anosmia, 147950	AD
	Jackson-Weiss syndrome, 123150	AD
	Osteoglophonic dysplasia, 166250	AD
	Pfeiffer syndrome, 101600	AD
	Trigonocephaly 1, 190440	AD
FGFR2	Antley-Bixler syndrome without genital anomalies or disordered steroidogenesis, 207410	AD
	Apert syndrome, 101200	AD
	Beare-Stevenson cutis gyrata syndrome, 123790	AD
	Bent bone dysplasia syndrome, 614592	AD
	Craniofacial-skeletal-dermatologic dysplasia, 101600	AD
	Craniosynostosis, nonspecific	
	Crouzon syndrome, 123500	AD
	Gastric cancer, somatic, 613659	
	Jackson-Weiss syndrome, 123150	AD
	LADD syndrome, 149730	AD
	Pfeiffer syndrome, 101600	AD
	Saethre-Chotzen syndrome, 101400	AD
	Scaphocephaly and Axenfeld-Rieger anomaly	
	Scaphocephaly, maxillary retrusion, and mental retardation, 609579	

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FGFR3	Achondroplasia, 100800	AD
	Bladder cancer, somatic, 109800	
	CATSHL syndrome, 610474	AR, AD
	Cervical cancer, somatic, 603956	
	Colorectal cancer, somatic, 114500	
	Crouzon syndrome with acanthosis nigricans, 612247	AD
	Hypochondroplasia, 146000	AD
	LADD syndrome, 149730	AD
	Muenke syndrome, 602849	AD
	Nevus, epidermal, somatic, 162900	
	SADDAN, 616482	AD
	Spermatocytic seminoma, somatic, 273300	
	Thanatophoric dysplasia, type I, 187600	
	Thanatophoric dysplasia, type II, 187601	AD
FREM1	Bifid nose with or without anorectal and renal anomalies, 608980	AD
	Manitoba oculotrichoanal syndrome, 248450	AR
	Trigonocephaly 2, 614485	AD
GLI3	Greig cephalopolysyndactyly syndrome, 175700	AD
	Pallister-Hall syndrome, 146510	AD
	Polydactyly, postaxial, types A1 and B, 174200	AD
	Polydactyly, preaxial, type IV, 174700	AD
	{Hypothalamic hamartomas, somatic}, 241800	
IFT122	Cranioectodermal dysplasia 1, 218330	
IL11RA	Craniosynostosis and dental anomalies, 614188	AR
KMT2D	Kabuki syndrome 1, 147920	AD
MASP1	3MC syndrome 1, 257920	AR
MEGF8	Carpenter syndrome 2, 614976	AR
MSX2	Craniosynostosis, type 2, 604757	AD
	Parietal foramina 1, 168500	AD
	Parietal foramina with cleidocranial dysplasia, 168550	AD
P4HB	Cole-Carpenter syndrome 1, 112240	AD
POR	Antley-Bixler syndrome with genital anomalies and disordered steroidogenesis, 201750	AR
	Disordered steroidogenesis due to cytochrome P450 oxidoreductase	AR
RAB23	Carpenter syndrome, 201000	AR
RECQL4	Baller-Gerold syndrome, 218600	AR
	RAPADILINO syndrome, 266280	AR
	Rothmund-Thomson syndrom, 268400	AR
SEC24D	Cole-Carpenter syndrome 2, 616294	AR
SKI	Shprintzen-Goldberg syndrome, 182212	AD
SMAD6	{Craniosynostosis 7, susceptibility to}, 617439	AD
TCF12	Craniosynostosis 3, 615314	AD

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TWIST1	Craniosynostosis 1, 123100	AD
	Robinow-Sorauf syndrome, 180750	AD
	Saethre-Chotzen syndrome with or without eyelid anomalies, 101400	AD
	Sweeney-Cox syndrome, 617746	AD
WDR35	Cranioectodermal dysplasia 2, 613610	AR
	Short-rib thoracic dysplasia 7 with or without polydactyly, 614091	AR
ZIC1	Craniosynostosis 6 , 616602	AD
ZNF462	Weiss-Kruszka syndrome, 618619	AD

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