Panelinformation Kraniosynostos in-silico

Version: 1.0

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	Transkript*	% täckning > 10x **	% täckning > 20x**	x Median täckning **	Associerad fenotyp, MIM# (OMIM)	Nedärv-ning
ALX4	NM_021926.3	100	100	152	Frontonasal dysplasia 2, 613451	AR
					Parietal foramina 2, 609597	AD
					{Craniosynostosis 5, susceptibility to}, 615529	AD
ASXL1	NM_015338.5	100	99	166	Bohring-Opitz syndrome, 605039	AD
					Myelodysplastic syndrome, somatic, 614286	
BMP4	NM_001202.5	100	100	174	Microphthalmia, syndromic 6, 607932	AD
					Orofacial cleft 11, 600625	
CD96	NM_198196.2	100	95	82	C syndrome, 211750	AD
CDC45	NM_001178010.2	100	100	158	Meier-Gorlin syndrome 7, 617063	AR
COLEC11	NM_024027.4	100	100	157	3MC syndrome 2, 265050	AR
CYP26B1	NM_019885.3	100	100	212	Craniosynostosis with radiohumeral fusions and other skeletal and craniofacial anomalies, 614416	
EFNB1	NM_004429.4	100	100	154	Craniofrontonasal dysplasia, 304110	XLD
ERF	NM_006494.3	100	100	145	Chitayat syndrome, 617180	AD
					Craniosynostosis 4, 600775	AD
FGFR1	NM_023110.2	100	100	159	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

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FGFR2	NM_000141.4	100	99	87	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	
FGFR3	NM_000142.4	100	100	128	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

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FREM1	NM_144966.5	100	99	98	Bifid nose with or without anorectal and renal anomalies, 608980 Manitoba oculotrichoanal syndrome, 248450 Trigonocephaly 2, 614485	AD AR AD
GLI3	NM_000168.5	100	100	177	Greig cephalopolysyndactyly syndrome, 175700 Pallister-Hall syndrome, 146510 Polydactyly, postaxial, types A1 and B, 174200 Polydactyly, preaxial, type IV, 174700 {Hypothalamic hamartomas, somatic}, 241800	AD AD AD AD
IFT122	NM_052985.3	100	100	154	Cranioectodermal dysplasia 1, 218330	
IL11RA	NM_001142784.2	100	100	164	Craniosynostosis and dental anomalies, 614188	AR
KMT2D	NM_003482.3	100	100	192	Kabuki syndrome 1, 147920	AD
MASP1	NM_001879.5	100	100	140	3MC syndrome 1, 257920	AR
MEGF8	NM_001410.2	100	100	120	Carpenter syndrome 2, 614976	AR
MSX2	NM_002449.4	100	100	127	Craniosynostosis, type 2, 604757 Parietal foramina 1, 168500 Parietal foramina with cleidocranial dysplasia, 168550	AD AD AD
POR	NM_000941.2	100	100	160	Antley-Bixler syndrome with genital anomalies and disordered steroidogenesis, 201750 Disordered steroidogenesis due to cytochrome P450 oxidoreductase	AR AR
RAB23	NM_016277.4	100	91	53	Carpenter syndrome, 201000	AR
RECQL4	NM_004260.3	100	100	181	Baller-Gerold syndrome, 218600 RAPADILINO syndrome, 266280 Rothmund-Thomson syndrom, 268400	AR AR AR
SKI	NM_003036.3	100	99	109	Shprintzen-Goldberg syndrome, 182212	AD
SMAD6	NM_005585.4	100	98	73	{Craniosynostosis 7, susceptibility to}, 617439	AD
TCF12	NM_207037.1	100	100	67	Craniosynostosis 3, 615314	AD

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TWIST1	NM_000474.3	100	97	123	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	NM_001006657.1	100	90	46	Cranioectodermal dysplasia 2, 613610	AR
					Short-rib thoracic dysplasia 7 with or without polydactyly, 614091	AR
ZIC1	NM_003412.3	100	98	240	Craniosynostosis 6 , 616602	AD

* Referenstranskript enligt NCBI Reference Sequences (RefSeq). Detta transkriptnummer används som utgångspunkt för analys av horisontell täckning. Däremot görs varianttolkning från genens hela kodande sekvens, oberoende av isoform.
** Medelvärde av 32 valideringsprov

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