SNOMED Concepts Associated with Difficult Airways (mostly Pediatric)

8Project proposed by Jorge Galvez

Objective

Incorporate list from Pedi-R group of SPA into a SNOMED CT refset, or perhaps a parent concept.

A thought - we could list specific SNOMED concepts, and for some concepts mark them as "also include all children." Then from that list we generate a flattened final refset.

Difficult Airway Finding

718446005 |Difficulty with mask ventilation and tracheal intubation (finding)|

Parent Concepts to Consider

Are all the children of these concepts indicators of difficult airways? Some of them?

Parent Concept	Notes
65094009 |Multiple malformation syndrome with facial defects as major feature (disorder)|	
77701002 |Multiple malformation syndrome, moderate short stature, facial (disorder)|	
32003007 |Congenital anomaly of face bones (disorder)|	
268239009 |Congenital abnormality of skull and face bones (disorder)|	Parent of 32003007 |Congenital anomaly of face bones (disorder)|
282041002 |Congenital abnormality of oral cavity (disorder)|	Not every one of these is a difficult airway
270516002 |Congenital macroglossia (disorder)|	

Specific Disorders

Disorder / Synonyms	SCTID	YMC notes	Include All Children?	Notes
Acrocephalosyndactyly type I Apert Syndrome	205258009 |Acrocephalosyndactyly type I (disorder)|	Include additional children (similar to craniosynostosis /also malformation of head and possible difficult airway)? ▪ Saethre-Chotzen syndrome (disorder) 83015004 ▪ Summitt syndrome (disorder) 733606001	Y	Should we include all children of parent 268262006 "Acrocephalosyndactyly (disorder)"? Yes. Please include all children. • Acrocephalopolysyndactyly (disorder) 205260006 • Acrocephalosyndactyly type I (disorder) 205258009 • Acrocephalosyndactyly type V (disorder) 70410008 • Craniosynostosis Philadelphia type (disorder) 720818003 • Curry Jones syndrome (disorder) 720819006

Arthrogryposis Freeman Sheldon Syndrome	52616002 |Freeman-Sheldon syndrome (disorder)| 715216008 |Distal arthrogryposis type 2B (disorder)|	Include these children if specific to head/face? ▪ Camptodactyly with joint contracture and facial skeletal defect syndrome (disorder) 715986009 ▪ Contracture with ectodermal dysplasia and orofacial cleft syndrome (disorder) 720746006 ▪ Holoprosencephaly sequence with hypokinesia and congenital joint contracture syndrome (disorder) 716169009 ▪ Hydrocephalus with cleft palate and joint contracture syndrome (disorder) 718576001 ▪ Malignant hyperthermia with arthrogryposis and torticollis syndrome (disorder) 719398004 ▪ Neurogenic arthrogryposis multiplex congenital (disorder) 715316005 Include these children since associated with small chin? ▪ Lethal congenital contracure syndrome type 1 (disorder) 715418007 ▪ Lethal congenital contracure syndrome type 2 (disorder) 715419004 ▪ Lethal congenital contracure syndrome type 3 (disorder) 715420005	Y	111246005 |Arthrogryposis (disorder)| isn't specific to the face - most common arthrogryposis involves distal part of limbs Insteadweshouldusethemorespecificvariants here. Alsorequestswere submitted to improve the modeling.
Barakat Syndrome also: HDR syndrome 10HDR-bakarat 10p-barakat	None	Not sure why this syndrome is associated with difficult airway, but this is the SNOMED code (no children): ▪ Hypoparathyroidism, deafness, renal disease syndrome (disorder) 724282009		OMIM 146255: "Hypoparathyroidism, sensorineural deafness, and renal disease" ORPHA2237
Beckwith WeidemannSyndrome	81780002 |Beckwith-Wiedemann syndrome (disorder)|		Y	
CHARGE Association	47535005 |Coloboma, heart malformation, choanal atresia, retardation of growth and development, genital abnormalities, and ear malformations association (disorder)|	Include additional parent "Charge-like syndrome?" ▪ Cleft palate with coloboma of eye and deafness syndrome (disorder) 718574003	Y	
Chromosome 11p13 deletion syndrome WAGR syndrome	715215007 |Chromosome 11p13 deletion syndrome (disorder)|		Y	

Congenital High Airway Obstruction Syndrome (CHAOS)		Not sure how to deal with this one...		Sounds like this is too broad to add to SNOMED - sometimes CHAOS manifests as tracheal atresia, sometimes as laryngeal atresia, sometimes laryngeal stenosis. Consider the following references. Can we create CHAOS as a parent concept for conditions that lead to high upper airway obstruction in the developmental period? Many of these are identified by fetal ultrasonography/MR imaging. Examples: create a concept of CHAOS - Congenital high airway obstruction syndrome that links the following conditions: Congenital atresia of larynx (disorder) SCTID: 64981002 Congenital atresia of larynx (disorder) SCTID: 64981002 Congenital stenosis of trachea due to tracheal web (disorder) SCTID: 447811005 Cyst of larynx (disorder) SCTID: 195867000 Congenital atresia of trachea (disorder) SCTID: 53189005 Subglottic stenosis (disorder) SCTID: 22668006 Congenital atresia of glottis (disorder) SCTID: 52879001 Agenesis of larynx (disorder) SCTID: 204535000 Congenital absence of trachea (disorder) SCTID: 3987009 Hamid-Sowinska, A., et al. (2011). "Congenital high airway obstruction syndrome." Neuro Endocrinol Lett **32**(5): 623-626. Congenital high airway obstruction syndrome (CHAOS) is a very rare fetal malformation caused by obstruction of fetal airway because of laryngeal or tracheal atresia, subglottic stenosis, laryngeal cyst or laryngeal web. The prenatal diagnosis is inferred from secondary changes such as enlarged, hyperechogenic lungs,ascites and/or hydrops, flattened or everted diaphragms, dilated distal airways and mediastinal compression. There are onlyfew cases of long-term survival described inliterature. We present the case offetus with such secondary changes diagnosed during routine ultrasound evaluation in 20 weeks' gestation. There were no other abnormalities and thekariotype was normal. In 26 weeks' gestation fetal hydrops appeared and subsequent polyhydramnios occurred in 28 weeks' gestation. The patient was planned for EXIT procedure during laborin experienced in CHAOS cases center. In 29 weeks'gestation the premature rupture of membranes and regular uterine contractions occurred and we've performedcesarean section. A multidisciplinary team of neonatologists,laryngologists and pediatric surgeons made their efforts to save the newborn, but therewas complete laryngeal atresia and trachealagenesia and immediate tracheostomy was impossible. The most important about CHAOS areearly diagnosis, detailed fetal assessment and an adequate postnatal intervention for establishing fetal airways. Gilboa, Y., et al. (2009). "Early sonographic diagnosis of congenital high-airwayobstruction syndrome." Ultrasound Obstet Gynecol **33**(6): 731-733. Vidaeff, A. C., et al. (2007). "More or less CHAOS: case report and literature review suggesting the existence of a distinct subtype of congenital high airway obstruction syndrome." Ultrasound Obstet Gynecol **30**(1): 114-117. Congenital obstruction of the upper airway (CHAOS) is a rare, usually lethal abnormality. A literature review of 36 prenatally diagnosed cases of CHAOS and the analysis of our own case suggest the existence of a distinct subtype of CHAOS, raising important implications for diagnosis and management. Serial fetal ultrasound examinations at 17-23 weeks' gestation showed hyperechoic and enlarged lungs, mediastinal shift, flattened diaphragm,polyhydramnios and apparently fluid-filled esophagus, findings interpreted as bilateral cystic adenomatoid malformation Type III. Ultrasound findings normalized around 32 weeks. The diagnosis of CHAOS was made after birth at term by direct laryngoscopy prompted by ventilatory difficulties and failed attempts at intubation. A pinhole opening posterior to the cricoid cartilage allowed the passage of an endotracheal tube. Based on observations in our case and those of five similar cases in the literature, we describe for the first time a subtype of CHAOS that is characterized by minorpharyngotracheal or laryngotracheal communications and associated with a less severe natural history and even resolution of ultrasound findings. In spite of this, a high index of awareness should be maintained becauseresolution of ultrasound findings does not necessarily indicateresolution of underlying pathology. Hedrick, M. H., et al. (1994). "Congenital high airway obstruction syndrome (CHAOS): a potential for perinatal intervention." J Pediatr Surg **29**(2): 271-274. Congenital high airway obstruction syndrome (CHAOS) results in a predictable constellation of findings: large echogenic lungs, flattened or inverted diaphragms, dilated airways distal to the obstruction, and fetal ascites and /or hydrops. The authors report on four fetuses referred for evaluation. None of them survived. Postmortem evaluation showed that three fetuses had laryngeal atresia, and one had tracheal stenosis. Coexistent fetal anomalies were accurately diagnosed by ultrasound in three of the four patients. The finding of CHAOS on prenatal ultrasound examination is diagnostic of complete or near-complete obstruction of the fetal upper airway, most likely caused by laryngeal atresia. A greater understanding of the natural history of CHAOS may permit improved prenatal and perinatal management.

Congenital Hypothyroidism	190268003 |Congenital hypothyroidism (disorder)|	I would favor using congenital hypothyroidism with diffuse goiter And consider these children due to frequent goiter: ▪ Endemic cretinism (disorder) 75065003 ▪ Familial thyroid dyshormonogenesis (disorder) 718183003 ▪ Iodide oxidation defect (disorder) 52724003 ▪ Iodide transport defect (disorder) 22558005 ▪ Sporadic cretinism (disorder) 84781002 ▪ Thyroid hormone responsiveness defect (disorder) 50375007 Include these due to association with cleft palate? ▪ Bamforth Lazarus syndrome (disorder) 722375007 Include these children due to association with craniosynostosis? ▪ Obesity, colitis, hypothyroidism, cardiac hypertrophy, developmental delay syndrome (disorder) 722051004	Y	Does this condition require goiter to be a difficult airway? See also: 278503003 |Congenital hypothyroidism with diffuse goiter (disorder)|
Congenital lingual tumor	127229000 |Neoplasm of lingual tonsil (disorder)|	Include all children: ▪ Benign neoplasm of lingual tonsil (disorder) 92184003 ▪ Carcincoma in situ of lingual tonsil (disorder) 92642005 ▪ Malignant tumor of lingual tonsil (disorder) 363377003 with all children Carcinoma of lingual tonsil (disorder) 254423005; Primary malignant neoplasm of lingual tonsil (disorder) 93868009; Primary squamous cell carcinoma of lingual tonsil (disorder) 722673007; Secondary malignant neoplasm of lingual tonsil (disorder) 94379004] ▪ Neoplasm of uncertain behavior of lingual tonsil (disorder) 94908004	Y	Itprobably doesn't matter if the tumor is congenital to be a difficult airway.
Congenital temporomandibular joint dysfunction		Should we include all TMJ joint dysfunction due to concern for poor mouth opening? ▪ Temporomandibular joint-pain-dysfunction syndrome (disorder) 386207004		See Poveda-Roda review of TMJ tumors/pseudotumors 235119009 | Mandibular condyle aplasia (disorder) | 708669006 | Bifid mandibular condyle (disorder) | 444552001 | Hyperplasia of mandibular bone (disorder) | 126551000 | Neoplasm of mandible (disorder) | 126550004 | Neoplasm of maxilla (disorder) | 50603008 | Ankylosis of temporomandibular joint (disorder) |
Cornelia de Lange Syndrome	40354009 |De Lange syndrome (disorder)|		Y	
Cri-Du-Chat	70173007 |5p partial monosomy syndrome (disorder)|	There is a Cri Du chat (finding) but I don't think that's associated with craniofacial abnormalities? 42712003	Y	
Cystic Hygroma	399882002 |Cystic hygroma (disorder)|	Include additional parent? ▪ Cystic lymphangioma (morphologic abnormality) 40225001	Y	

DiGeorge Sequence (22Q Deletion)	77128003 |DiGeorge sequence (disorder)|	Not listed under DiGeorge sequence: ▪ Deletion of part of chromosome 22 (disorder) 726399005	Y	There are several variants of 22q deletion Most common is22q11.2 deletion which is associated with both DiGeorge syndrome and velocardiofacial syndrome (below). See http://www.omim.org/entry/192430 Many of the terms have the same parent, "DiGeorge Sequence" so I think that might be adequate.
Down Syndrome	41040004 |Complete trisomy 21 syndrome (disorder)|	Include additional parents? ▪ Translocation Down syndrome (disorder) 371045000 ▪ Trisomy 21-mitotic nondisjunction mosaicism (disorder) 205616004 – maybe not this one since it should be a milder form of Down Syndrome?	Y	
Emanuel Syndrome 11:22 chromosomal translocation	702417004 |Supernumerary der(22)t(11;22) syndrome (disorder)|		Y	OMIM 609029 ORPHA96170
Emery Dreifuss Muscular Dystrophy	111508004 |Emery-Dreifuss muscular dystrophy (disorder)|		Y	
Epidermolysis Bullosa	61003004 |Epidermolysis bullosa (disorder)|	Not sure why this is a possible difficult airway...	Y	
Escobar Syndrome	80773006 |Escobar syndrome (disorder)|			
Fibrodysplasia Ossificans Progressiva Syndrome	82725007 |Progressive myositis ossificans (disorder)|			
First Arch Syndrome	15557005 |First arch syndrome (disorder)|	Include additional parent? (Japanese case report: Airway obstruction after general anesthesia in a patient with the first and second branchial arch syndrome. Masui. 2000 Nov;49 (11):1270-3) ▪ First and second branchial arch syndrome (703973009)	Y	Child of megaparent 65094009
Goldenhar Hemifacial Microsomia	205418005 |Goldenhar syndrome (disorder)|		Y	
Hunter Syndrome	70737009 |Mucopolysaccharidosis type II (disorder)|	Include additional? ▪ Trigonocephaly with broad thumb syndrome (disorder) 719949001 aka Hunter Rodd Hoffman syndrome	Y	
Hurler Syndrome	65327002 |Mucopolysaccharidosis type I-H (disorder)|	Include additional? ▪ Mucopolysaccharidosis type I-H/S (disorder) 26734009 aka Hurler-Scheie	Y	
Hunter-Mcalpine Craniosynostosis Syndrome	None	▪ Hunter McAlpine craniosynostosis syndrome (disorder) 721227001		Need to submit request to add concept OMIM 601379 ORPHA97340
Klippel-Feil Syndrome	5601008 |Klippel-Feil sequence (disorder)|	Include additional? ▪ Congenital dystrophia brevicollis (disorder) 388981000 aka Bonnevie-Ullrich and Klippel-Feil syndrome	Y	

Laryngeal Cleft		Parent: ■ Congenital cleft larynx (disorder) 232461002 has children: -Congenital cleft of posterior cricoid cartilage (disorder) 204558002 -Laryngeal cleft type I (disorder) 306949002 -Laryngeal cleft type II (disorder) 306950002 -Laryngeal cleft type III (disorder) 306951003 -Laryngeal cleft type IV (disorder) 306953000 -Opitz Frias syndrome (disorder) 81771002		many types...
Laryngeal Web	297159008 |Laryngeal web (disorder)|	Children: ■ Acquired laryngeal web (disorder) 232447007 ■ Congenital web of larynx (disorder) 47070001 ■ Subglottic web (disorder) 444921008	Y	Include all children?
Laryngeal Hemangioma	703199001 |Laryngotracheal hemangioma (disorder)|	Shouldn't "subglottic hemangioma" and "laryngeal hemangioma" be considered children of the parent "laryngotracheal hemangioma?" Include additional parents? ■ PHACE Posterior fossa brain malformation, hemangioma, arterial anomaly, cardiac defect and aortic coarctation, and eye abnormality syndrome (disorder) 698765007 ■ PHACES Posterior fossa brain malformation, haemaniogma, arterial anomaly, cardiac defect and aortic coarctation, eye abnormality syndrome and sternal anomaly syndrome (disorder) 698766008	Y	Should we add synonyms "subglottic hemangioma" and "laryngeal hemangioma" to this term? Or are those separate concepts? Should this concept be added as a child of 60600009 | Disorder of the larynx (disorder)| ?
Li-Fraumeni Syndrome	428850001 |Li-Fraumeni syndrome (disorder)|		Y	
Lipoid Proteinosis	38692000 |Lipid proteinosis (disorder)|		Y	
Microstomia	14582003 |Microstomia (disorder)|		Y	
Moebius Syndrome	429753001 |Congenital nonprogressive myopathy with Moebius and Robin sequences (disorder)|	Include additional parent? ■ Moebius syndrome, axonal neuropathy, hypogonadotropic hypogonadism syndrome (disorder) 724174003	Y	Another 65094009 child

Neurofibromatosis Type 1	81669005 |Neurofibromatosis (morphologic abnormality)|	NF1 patients have been reported, though rarely, to have fibromas in the oropharynx (e.g., tongue base, larynx). NF2 is more rare, but a case report of difficult airway exists: ▪ Haldar R, Khandelwal A, Vagyannavar R, Srivastava S (2017) Obscure retropharyngeal mucocutaneous masses associated with acoustic neurofibromatosis: A source of difficult airway management. J Neurosurg Anesthesiol. 29(3):369-370. Interestingly, both NF1 and NF2 are associated with cervical lesions that pose a special concern for intubation (e.g., developemtn of hematoma with neck extension or jaw thrust maneuver)	Y	Do most patients with NF have a difficult airway? Both Type 1 and Type 2?
Noonan's Syndrome	205824006 |Noonan's syndrome (disorder)|		Y	
Prader WilliSyndrome	89392001 |Prader-Willi syndrome (disorder)|	Include additional parents?: ▪ Royer's syndrome (disorder) 37355009 aka Prader Willi syndrome AND diabetes ▪ Intellectual disability and short stature with hand contracture and genital anomaly syndrome (disorder) 716334004 aka Prader Willi habitus with osteopenia and camptodactyly	Y	
Rheumatoid Arthritis		If limiting this to RA of cervical spine, then perhaps also include?: ▪ Rheumatoid arthritis of temporomandibular joint (disorder) 427770001		Parent term 69896004 is pretty broad Should we only include 201764007 | Rheumatoid arthritis of cervical spine (disorder) | 69896004 |Rheumatoid arthritis (disorder)|
Robin Sequence	4602007 |Robin sequence (disorder)|	Include additional (not listed as children under Robin sequence)?: ▪ Pierre Robin sequence, congenital heart defect, talipes syndrome (disorder) 725911008 aka TARP syndrome ▪ Congenital nonprogressive myopathy with Moebius and Robin sequences (disorder) 429753001	Y	
Rubinstein-Taybi Syndrome	45582004 |Rubinstein-Taybi syndrome (disorder)|		Y	
Smith-Lemli-Opitz syndrome (disorder)	43929004 |Smith-Lemli-Opitz syndrome (disorder)|			Child of 77701002 | Multiplemalformationsyndrome, moderate short stature, facial (disorder) |
Stickler Syndrome	78675000 |Stickler syndrome (disorder)|			
Tracheal Stenosis	11296007 |Stenosis of trachea (disorder)|	I would favor including all children		Include all children?
Treacher-Collins	82203000 |Treacher Collins syndrome (disorder)|			Child of big parent 65094009

Trisomy 4p	49024004 |4p partial trisomy syndrome (disorder)|	Include additional?: ■ 4p16.3 microduplication syndrome (disorder) 726706008 aka Distal trisomy 4p (or not since dysmorphic features described do not necessarily seem like difficult airway concerns – "high forehead with frontal bossing, hypertelorism, prominent glabella, long narrow palpebral fissures, low set ears, and short neck."		
Trisomy 8	205649008 |Trisomy 8 (disorder)|	Interestingly, "Trisomy 8" only has parent "Trisomy and partial trisomy of autosome" There is another parent "Anomaly of chromosome pair 8 (disorder) 48082007" but it includes some minor mutations in chromosome 8 that may not manifest in severe enough symptoms to warrant difficult airway. If not including all children in 48082007, then perhaps include: ■ Complete trisomy 8 syndrome (disorder) 68454002 ■ Deletion of part of chromosome 8 (disorder) 726378007 - case report in Korean J Anesthesiol 2011 61(4) describing difficult intubation due to macroglossia, short neck, high arched palate ■ Mosaic trisomy 8 syndrome (disorder) 717335009		
Trisomy 9	205650008 |Trisomy 9 (disorder)|	Similar to trisomy 8, "Trisomy 9" only has parent "Trisomy and partial trisomy of autosome." There are additional parents: ■ Partial trisomy of chromosome 9 (disorder) 726348003 ■ Anomaly of chromosome pair 9 (disorder) 5051002 There is a case report of laryngeal atresia with partial trisomy (Genet Couns 1991 2 (2):83-91), but if not interested in partial trisomy, note there is another entry under 5051002 (similar to trisomy 8 above) ■ Complete trisomy 9 syndrome (disorder) 74350000		
Trisomy 13 (Patau Syndrome)	254268004 |Partial trisomy 13 in Patau's syndrome (disorder)| 21111006 |Complete trisomy 13 syndrome (disorder)|			

Trisomy 18 (Edwards Syndrome)	51500006 |Complete trisomy 18 syndrome (disorder)|	Consider "Anomaly of chromosome pair 18 (disorder) 59033006?" Includes children: ▪ Complete trisomy 18 syndrome (disorder) 51500006 ▪ Deletion of part of chromosome 18 (disorder) 726391008 ▪ Partial trisomy 18 in Edward's syndrome (disorder) 254266000 ▪ Partial trisomy of chromosome 18 (disorder) 726357009 ▪ Ring chromosome 18 syndrome (disorder) 88154004 ▪ Tetrasomy 18p (disorder) 698849002		
Trisomy 22	205655003 |Trisomy 22 (disorder)|	Similar to trisomy 8 and 9, "Trisomy 22" only has parent "Trisomy and partial trisomy of autosome." Include additional: ▪ Complete trisomy 22 syndrome (disorder) 71703005 ▪ Partial trisomy of chromosome 22 (disorder) 726362005		
VACTERL	431395004 |Vertebral abnormalities, anal atresia, cardiac abnormalities, tracheo-esophageal fistula, renal anomalies, limb defects syndrome (disorder)|	Include additional: ▪ Vertebral abnormality, anal atresia, cardiac abnormality, tracheo-esophageal fistula, renal anomaly, limb defect syndrome with hydrocephalus (disorder) 719043002 aka VACTERL with hydrocephalus		
Vallecular Cyst	232410007 |Vallecular cyst (disorder)|			
Velocardiofacial Syndrome (Shprintzen Syndrome)	83092002 |Shprintzen syndrome (disorder)|			
Weaver Syndrome	63119004 |Weaver syndrome (disorder)|	Include additional: ▪ Weaver Williams syndrome (disorder) 726670008		

Terminology Requests

Request Type	Details	CRS Request ID & Status

Add Child	83015004 |Saethre-Chotzen syndrome (disorder)| should be child of 268262006 |Acrocephalosyndactyly (disorder)| Note that "Acrocephalosyndactyly, type V" is already a synonym. Compare concepts	CRS 444115 Ready for Release
Add Child	28861008 |Crouzon syndrome (disorder)| should be child of 268262006 |Acrocephalosyndactyly (disorder)| Note that "Acrocephalosyndactyly, type II" is already a synonym. Compare concepts	CRS 718445 Pending Clarification: "28861008|Crouzon syndrome (disorder)| is currently a descendant of 57219006|Craniosynostosis syndrome (disorder)| and this aligns with Orphanet and the ICD-11 draft. Proposal: inactivate the description Acrocephalosyndactyly, type II as there does not appear to be evidence that this is a current synonym for the disease." Note that OMIM does Acrocephalosyndactyly type II as a synonym, but for Apert Syndrome (which is confusing). Also see Bissonnette B, Dalens BJ. Syndromes, rapid recognition and perioperative implications. McGraw-Hill Professional. (2006) ISBN:0071354557.
Rename / Add Synonym	52616002 |Freeman-Sheldon syndrome (disorder)| should be renamed to "Distal arthrogryposis type 2A (disorder)" with the original name as a synonym. See OMIM 193700 for reference	CRS 718446 Merged with below
Add Child	52616002 |Freeman-Sheldon syndrome (disorder)| should be a child of 24269006 |Distal arthrogryposis syndrome (disorder)| See OMIM 193700 for reference	CRS 718447 The concept 52616002|Freeman-Sheldon syndrome (disorder)| has been remodelled to become a child of 24269006|Distal arthrogryposis syndrome (disorder)|. A new synonym has been added Distal arthrogryposis type 2A and a text definition has been added to the concept.

Add Concept	FSN: Hypoparathyroidism-deafness-renal disease syndrome Description: Barakat syndrome HDR syndrome OMIM 146255: "Hypoparathyroidism, sensorineural deafness, and renal disease" ORPHA2237	CRS 719796 Proposed parent is 74345006 |Congenital disorder due to abnormality of chromosome number OR structure (disorder)|
Add Concept	Hunter-Mcalpine Craniosynostosis Syndrome OMIM 601379 ORPHA97340	CRS 719797 Proposed parent is 57219006 |Craniosynostosis syndrome (disorder)|