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| **Supplemental Table 1A. Overview of disorders characterized by co-occurrence of craniosynostosis and congenital diaphragmatic hernia** **Clinical disorders with autosomal dominant inheritance pattern** |  |
|  | **Gene****(MIM number)** | **PhenotypeMIM number** | **chromosome** | **CS\*** | **CDH‡** |  **Key clinical features** | **Authors reporting on CDH and/or CS** |
| **Apert syndrome**  | FGFR2 (176943) | 101200 | 10q26.13 | Key feature, multisutural, progressive | Rare (six cases of CDH (1-6) and one case of diaphragm agenesis(7)) | * Symmetric syndactyly of hands and feet
* Midface hypoplasia(8)
 | Kaur2019Dap2019Kosinski2016Sobaih2015Bulfamante2011Wallis-Crespo2004Witters2000 |
| **Kabuki syndrome (focus on type 1)** | KMT2D (602113) | 147920 | 12q13.12 | Occasional(9-11) | Relatively common(12-15) | * Characteristic facial features: long palpebral fissures, everted lower eyelids, ptosis, arched eyebrows, blue sclera, cupped ears, micrognathia
* Short stature, microcephaly
* Intellectual disability (mild to moderate)
* High/cleft palate and dental anomalies
* Brachydactyly, clinodactyly, persistent fetal pads
* Cardiac anomalies (16)
 | Scott2021Topa2017Martinez-Lopez2010David2004Geneviève2004Van Haelst2000 |
| **CEBALID\*\* syndrome** **(MN1 C-terminal truncation syndrome)** | MN1 (156100) | 618774 | 22q12.1  | Reported in three patients out of 25 identified patients identified to date(17, 18) | Reported in two patients out of 25 identified patients identified to date (17, 18)  | * Characteristic facial features: midface hypoplasia, downslanting palpebral fissures, hypertelorism, exophthalmia, low-set ears, a short upturned nose
* Intellectual disability, hypotonia, delay in motor development
* Hearing loss
* Structural brain anomalies(17, 18)
 | Mak 2020 |
| **Chromosome 22q11.2 deletion syndrome** |  | 145410; 188400; 192430; 600594; 601279; 601755; 602054; 609030 | 22q11.2 | Rare feature(19) (*may include CDC45 pathogenic variant in remaining allele*(20)) | Rare feature(21)  | * Highly variable phenotype (ranging from minor abnormities to major structural defects)
* Cardiovascular anomalies
* Cleft palate
* Cognitive impairment
* Short stature
* Characteristic facial features: hypoplastic nasal alae, wide nasal bridge, short palpebral fissures,

low-set, small ears * Nasal speech(22, 23)
 | Unolt 2020, 2017McDonal-McGinn 2005 |
| **SPECC1L- related syndromes**  | SPECC1L (614140) | 145410145420 600251  | 22q11.23 | Occasional(24)  | Occasional occurrence(24-27) | * Characteristic facial features: hypertelorism, a wide, short nose, ptosis and retrognathia
* Cleft lip/palate
* Clinical features include branchial fistulas, omphalocele, genitourinary anomalies(28)
 | Wild 2020Bhoj 2019Kruszka 2015Robin 1995 |
| **7q11. 23 Duplication syndrome** | - | 609757 | 7q11.23 | Rare(29-31) | Rare(29, 32) | * Variable expression, with incomplete penetrance
* Characteristic facial features: prominent forehead, hypertelorism, high and broad nose, straight eyebrows, and thin lips
* Cognitive impairment and intellectual disability
* Epilepsy(33)
 | Morris 2015Van der Aa 2009Torniero 2008Kriek 2006 |
| **X-Linked**  |  |  |  |  |  |
| **Craniofrontonasal syndrome (XLD)** | EFNB1 (300035) | 304110 | Xq13.1  | Common feature, often either unilateral or bilateral coronal CS(34, 35) | Relatively common/ occasional(36-44) | * More severe phenotype in females
* Characteristic facial features: hypertelorism, craniofacial asymmetry, webbed neck, bifid tip of the nose, a broad nasal bridge
* clinodactyly of ≥ 1 digit
* longitudinal splitting/ridging of nails(35)
 | Hogue 2010Kawamoto 2007Vasudevan 2006Twigg 2004 & 2006Brooks 2002McGaughran2002Hurst 1988Morris 1987 |
| **Cornelia de Lange syndrome**  | NIPBL (608667, AD)SMC1A(300040, XLD) | 122470300590 | 5p13.2 Xp11.22 | Described for NIPBL variant(45); and for SMCA1(46) | Key feature (47) | * Characteristic facial features: thick, arched eyebrows or synophrys, long/smooth philtrum, short nose, thin upper vermillion
* Limb defects
* Intellectual disability
* Growth retardation
* Hirsutism (48)
 | Desai 2021Xu 2018Gupta 2020 |
| **Simpson-Golabi-Behmel syndrome, Type 1****(XLR)** | GPC3(300037) | 312870 | Xq26.2  | 3 case reports(49-51) | Occasional(49, 52)  | * Characteristic facial features: hypertelorism, downslanting palpebral fissures
* Cleft palate/lip.
* Overgrowth and macrocephaly
* Intellectual disability
* Cardiac anomalies
* Renal abnormality,
* Brachy-, syn-, and polydactyly.(53)
 | Schirwani 2019Villarreal 2013Li 2009 |

**Supplemental Table 1A . Overview of disorders characterized by co-occurrence of craniosynostosis and congenital diaphragmatic hernia.** This table is the supplemental version of Table 2A in the article. It presents an overview of clinical disorders in which both craniosynostosis and congenital diaphragmatic hernia have been reported more than once. Abbreviations: \*CS= craniosynostosis, ‡CDH = congenital diaphragmatic hernia, \*\* CEBALID= craniofacial defects, dysmorphic ears, structural brain abnormalities, expressive language delay, and impaired intellectual development, XLD= X-linked dominant, XLR= X-linked recessive.

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| **Supplemental Table 1B. Isolated case reports on the co-occurrence of craniosynostosis and congenital diaphragmatic hernia** |  |
|  | **Gene****(MIM number)** | **PhenotypeMIM number** | **chromosome** | **CS\*** | **CDH‡** |  **Key clinical features** | **Authors reporting on CDH and/or CS** |
| **Saethre -Chotzen** | TWIST1(601622) | 101400 | 7p21.1  | Key feature, often bicoronal CS | One case(54), unclear if co-occurrence of CDH is coincidental. Mouse models suggest a possible role for TWIST1 in development of the diaphragm | * Characteristic facial features: ptosis, downward slanting palpebral fissure, depressed nasal bridge, facial asymmetry
* Small ears with prominent crus
* Syndactyly of hand and feet (55)
 | Piard 2012 |
| **Chromosome 9p deletion syndrome** | - | 158170 | 9p | Key feature: metopic CS | One case described for 9p deletion syndrome(56)  | * Characteristic facial features: hypotelorism, upslanting palpebral fissures, low-set ears, malformed ears, long philtrum
* Moderate to severe intellectual disability
 | Alfi 1973 |
| **15q24 deletion syndrome** | - | 613406 | 15q24.2 | 1 case report(57) | Four reports (58-60) | * Characteristic facial features: high forehead, facial asymmetry, downslanting of eyes, hypertelorism, and a long smooth philtrum, ear malformations
* Intellectual disability
* Genitourinary anomalies
* Cardiovascular malformations(61, 62)
 | Ng 2011Van Esch 2009Sharp 2007Bettelheim 1998 |
| **DPF2-related Coffin–Siris syndrome** | DPF2(601671) | 618027 | 11q12.1 | At least two out of a total of 10 reported patients (one patient was stated to have trigonocephaly but no x-ray was performed)(63) | One patient described out of a total of 10 reported patients.(64)  | * Cognitive impairment, intellectual disability, and behavioral problems
* Feeding problems and hypotonia
* Hearing loss
* Brachydactyly, clinodactyly, hypoplastic nails
* Coarse facial features
 | Knapp 2019Vasileiou 2018 |
| - | DSC2(125645) | - |  | One report of a patient with multisutural CS and CDH (65) | One report of a patient with multisutural CS and CDH (65) | Isolated case: presented with left atrial isomerism, transposed systemic and pulmonary veins, intestinal malrotation, bilateral inguinal hernia, hydronephrosis and nephrolithiasis in addition to CDH and CS (65) | Das 2019 |
| **Loeys-Dietz syndrome**  | TGFBR1 (190181) TGFBR2(190182) | 609192610168 | 9q22.333p24.1 | Multiple cases reported (66) | One report(67) | * Aortic and arterial aneurysms
* Characterstic facial features: hypertelorism, downslant of the eyes
* Cleft palate, bifid uvula.
* Pectus anomalies
* Arachnodactyly (66)
 | Lobaton 2021Loeys 2005 |
| **Gain of function of RARB** | RARB |  |  | One report of a patient with CS(68) | Multiple patients with diaphragmatic hernia(68, 69) | Thirteen cases have been reported in total. Clinical features include microphthalmia and anophthalmia, sclerocornea, and coloboma, as well as cardiac anomalies, and malrotation of the bowel (68, 69) | Srour 2016 |

**Supplemental Table 1B. Isolated case reports on the co-occurrence of craniosynostosis and congenital diaphragmatic hernia.** This table is the supplemental version of Table 2B in the article.Abbreviations: \*CS = craniosynostosis, ‡CDH = congenital diaphragmatic hernia.

Mutations in FRAS-related extracellular matrix protein 1 (FREM1, MIM number: 608944) are not shown in this table as reports on this gene are contradictory. Vissers et al suggested CS is a feature of heterozygous FREM1 mutations,(70) although this finding was not supported by Dawson et al (71). CDH, in contrast, has been reported in a patient with a recessive FREM1mutation.The effect of the latter was supported by a mouse model.(72) Similarly, there have been somewhat conflicting reports on craniosynostosis in Wolf-Hirschorn syndrome, which includes deletions of 4p16.1. The effect of deletion of FGFRL1, located on 4p16.3 has been implicated in the development of craniosynostosis in both mouse models and humans.(73) In addition, a case report described a patient with a de novo 4p15 deletion, who presented with metopic synostosis.(74) However, the characteristic 4p16.1 was not included in the deletion and therefore this patient was considered to have a different phenotype.(74) In contrast, there have been several reports on patients with Wolf-Hirschorn who presented with CDH.(75-77)

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