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Evaluation of ICD-9-CM Codes for Craniofacial Microsomia

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Abstract

Background—Craniofacial microsomia (CFM) is a congenital condition characterized by microtia and mandibular underdevelopment. Healthcare databases and birth defects surveillance programs could be used to improve knowledge of CFM. However, no specific ICD-9-CM code exists for this condition, which makes standardized data collection challenging. Our aim was to evaluate the validity of existing ICD-9-CM codes to identify individuals with CFM.

Methods—Study sample eligibility criteria were developed by an expert panel and matched to 11 ICD-9-CM codes. We queried hospital discharge data from two craniofacial centers and identified a total of 12,254 individuals who had ≥ 1 potentially CFM-related code(s). We reviewed all (n=799) medical records identified at the University of North Carolina (UNC) and 500 randomly selected records at Seattle Children's Hospital (SCH). Individuals were classified as a CFM case or non-case.

Results—Thirty-two individuals (6%) at SCH and 93 (12%) at UNC met the CFM eligibility criteria. At both centers, 59% of cases and 95% of non-cases had only one code assigned. At both centers, the most frequent codes were 744.23 (microtia), 754.0 and 756.0 (nonspecific codes), and the code 744.23 had a positive predictive value (PPV) >80% and sensitivity >70%. The code 754.0 had a sensitivity of 3% (PPV<1%) at SCH and 36% (PPV=5%) at UNC, whereas 756.0 had a sensitivity of 38% (PPV=5%) at SCH and 18% (PPV=26%) at UNC.

Conclusions—These findings suggest the need for a specific CFM code to facilitate CFM surveillance and research.

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Keywords

craniofacial microsomia; hemifacial microsomia; oculo-auriculo-vertebral spectrum; microtia; Goldenhar; birth defects surveillance; ICD-9-CM

Introduction

Craniofacial microsomia (CFM) is a congenital condition characterized by underdevelopment of the ear (microtia and anotia) and mandible, and often includes extracranial anomalies (Gorlin et al., 2001). Wide phenotypic variation is characteristic of CFM, and microtia has been considered a mild form (Bennun et al., 1985; Grabb, 1965; Melnick, 1979; Rollnick and Kaye, 1983; Tasse et al., 2007). Though CFM is typically associated with facial asymmetry, bilateral involvement has been reported in a high proportion of individuals with CFM (Heike and Hing, 2009). The etiology of this condition is unknown, and CFM remains a clinical diagnosis, though there are no established clinical diagnostic criteria for CFM. We use the term CFM to encompass other terms that have been used interchangeably for the same phenotype, including hemifacial microsomia, Goldenhar syndrome, and oculo-auricular-vertebral spectrum. The estimated birth prevalence of this highly heterogeneous condition is 1 in 5,600 live births (Cohen et al., 1989; Grabb, 1965), making it the third most common congenital craniofacial condition. Craniofacial microsomia often affects breathing, hearing, dental occlusion and speech. According to published guidelines, children with CFM require longitudinal, multidisciplinary health care (Brent, 1999; Heike and Hing, 2009; Monahan et al., 2001). Despite its clinical importance in terms of frequency, impact on health and functioning, and burden of care, there are no *International Classification of Diseases, 9th Revision, Clinical Modification* (ICD-9-CM) codes specific to CFM.

Lack of such a code has been a barrier to effective surveillance of and research on CFM, resulting in a dearth of literature regarding CFM etiology, clinical course, health care services, and quality of life for individuals with this condition. With the use of a specific CFM code, these areas of inquiry could be explored through hospital discharge databases, national healthcare databases, and birth defects surveillance programs. Multicenter collaborations are needed to conduct rigorous epidemiologic studies regarding possible risk factors, yet such investigations require standardized case ascertainment procedures. Data collection standardization can be greatly enhanced by incorporating relevant and specific diagnostic codes.

Given the current lack of a specific CFM ICD-9-CM code and no plans to incorporate such a code in the new ICD-10, reaching consensus on the most applicable code(s) would be a first step to determine if any of the current ICD-9-CM codes are practical for CFM case ascertainment. In addition, determining specific codes for CFM would facilitate surveillance of and research on this condition. Our study objective was to estimate the sensitivity and positive predictive value of selected ICD-9-CM codes for the identification of individuals with CFM.

Methods

This study was conducted through a multicenter consortium entitled the “Facial Asymmetry Collaborative for Interdisciplinary Assessment and Learning (FACIAL).” The FACIAL network was supported by the National Institute of Dental and Craniofacial Research to address gaps in knowledge regarding the etiology and care for children with CFM. FACIAL presently consists of four U.S. craniofacial centers. The network’s first tasks were to

develop a consensus-based research definition of CFM and to delineate specific case eligibility criteria. The expert panel, convened for these purposes, included team members from several U.S. craniofacial centers and included: craniofacial pediatricians, dentists, epidemiologists, medical geneticists, oral and maxillofacial surgeons, orthodontists, otolaryngologists, and plastic and reconstructive surgeons with experience in the diagnosis and care of children with CFM.

The craniofacial features associated with CFM range from subtle mandibular asymmetry with a preauricular skin tag to more severe forms that include microtia, absence of the ear canal, facial clefts, and/or epibulbar dermoids. An exhaustive list of CFM features was compiled and discussed at an in-person FACIAL network meeting and the resulting consensus led to the eligibility criteria. The CFM case inclusion criteria were defined as at least one of the following: 1) microtia; 2) anotia; 3) facial asymmetry and preauricular tag; 4) facial asymmetry and facial tag; 5) facial asymmetry and epibulbar dermoid; 6) facial asymmetry and macrostomia; 7) preauricular tag and epibulbar dermoid; 8) preauricular tag and macrostomia; 9) facial tag and epibulbar dermoid; 10) macrostomia and epibulbar dermoid.. Clinical features of mandibular hypoplasia, soft tissue deficiency, facial nerve palsies, and/or orbital dystopia were categorized under facial asymmetry, unless individual had clear indication of symmetric involvement. Our consensus panel agreed that children with bilateral CFM are typically asymmetrically affected. Malformations associated with CFM can be seen in other, well-known syndromes and in individuals with chromosomal abnormalities. We excluded potential cases with a syndromic diagnosis (e.g. Treacher Collins syndrome) or chromosomal abnormality in order to restrict our study population to individuals with CFM of unknown etiology.

We generated an exhaustive list of ICD-9-CM codes that could potentially capture individuals with the CFM inclusion criteria. Nine ICD-9-CM codes specifically matched at least one inclusion criterion (Table 1). We also included two additional ICD-9-CM codes, 756.0 (“congenital anomalies of skull and face bones”) and 754.0 (“musculoskeletal deformities of skull face and jaw”), used by providers for patients with CFM, although these codes are less specific. Henceforth, we refer to all of these identified codes as “CFM codes.”

The current study was conducted at two craniofacial centers, Seattle Children’s Hospital Craniofacial Center (SCH) and University of North Carolina Craniofacial Center (UNC). To identify individuals who might carry a CFM diagnosis, we queried the inpatient and outpatient billing databases for the longest period available for electronic billing records through each institution: (1) SCH between October 1, 1997 to September 30, 2011 and (2) UNC between July 1, 2004 and January 18, 2011. Specifically, the queries identified any patient seen at either hospital who had received one or more of the CFM codes. This search yielded a total of 11,455 unique patients at SCH and 799 at UNC.

The main outcomes were the sensitivity and positive predictive value for ascertaining CFM by using the CFM codes, in isolation and in combination. We abstracted medical records from patients ascertained in this manner, reviewing all 799 medical records identified at UNC and 500 records randomly selected from the 11,455 patients identified at SCH using a formally designed and structured abstraction form. A trained healthcare professional (CLH and DV) from each center reviewed each medical chart at her respective institution and recorded any clinical descriptions that corresponded to the list of CFM eligibility criteria. We classified individuals as having a CFM diagnosis (case) or not (non-case) based on whether individuals met the eligibility criteria.

We then compared the CFM codes identified in the records of those who met the CFM eligibility criteria and those who did not. We summarized the distribution of number of

codes per individual. We also calculated the sensitivity and positive predictive values and corresponding 95% confidence intervals (CI) for each code and code combination. The sensitivity of each ICD-9-CM code was defined as the number of CFM cases who had the respective code in the billing data divided by the number of CFM cases overall. Positive predictive value was calculated as the proportion of individuals with a particular ICD-9-CM code who met CFM case criteria. We were unable to calculate the specificity and negative predictive value given a lack of information on the number of individuals in the respective databases without CFM codes and without a diagnosis of CFM.

The codes 743.8 and 744.21 were never used in the UNC data nor in the records selected for review at SCH. In order to assess the sensitivity of these codes for ascertaining CFM, we reviewed the medical records of the 31 individuals among the total 11,455 individuals with CFM codes at SCH that had these codes.

We performed all analyses in Stata version 10.1 (StataCorp, 2007). Institutional Review Board (IRB) approval was obtained for all study procedures at Seattle Children's Research Institute IRB and UNC Biomedical IRB.

Results

For the 500 individuals included in the medical chart abstraction at SCH, 58% were male. For the 799 individuals at UNC, 525 (64%) were male. The distribution of CFM codes was similar among sampled and non-sampled patients at SCH.

At UNC, 93 individuals (12%) met the CFM eligibility criteria, whereas 32 (6%) met the CFM eligibility criteria in the SCH sample. Overall, the codes most frequently identified included the two non-specific craniofacial codes, 754.0 and 756.0, and the code for microtia, 744.23 (Table 2). The codes for other specified congenital anomalies of the eye (743.8), congenital absence of external ear (744.01), accessory auricle (744.1), and congenital absence of ear lobe (744.21) were not identified in records at UNC. At SCH, codes 743.8 and 744.21 were also not identified in the sample of 500 records and in only 26 and 5, respectively, of the total 11,455 records queried at SCH. Review of charts for these 31 records revealed that only one individual (with 744.21 code) met the case eligibility criteria.

At both centers, 59% of cases had only one code assigned compared with over 95% of non-cases. The code for microtia (744.23) was frequently included in records of cases with two CFM codes. At SCH, the most common additional codes were those for "other congenital anomalies of external ear with impairment of hearing" (744.02) and anotia (744.01). At UNC, the most common additional codes were the non-specific 754.0 and 756.0 (Table 2).

The microtia code (744.23) was the most frequently identified code among cases with 72% and 82% at SCH and UNC, respectively. The positive predictive value for this code was above 80% in both centers. The code 754.0 had a sensitivity of 3% (95% CI: 0–16) at SCH and 35.5% (95% CI: 26–46) at UNC, whereas the code 756.0 had a sensitivity of 38% (95% CI: 21–56) at SCH and 18% (95% CI: 11–28) at UNC. Though 756.0 had a positive predictive value of 26% (95% CI: 16–39) at UNC, the positive predictive value for 756.0 at SCH was only 5% (95% CI: 3–9). Positive predictive values were less than or equal to 5% for 754.0 at both centers. At SCH, codes 744.01 and 744.1 had sensitivities of 19% (95% CI: 7–36) and 6% (95% CI: 1–21), respectively (Table 2).

The vast majority of non-cases had CFM codes 754.0 and/or 756.0. The diagnosis of skull deformity was the most common condition identified from medical charts of non-cases. Other diagnoses included: dysmorphic features, macrocephaly, known syndromes, and craniosynostosis. We identified 14 and 20 individuals at UNC and SCH, respectively, for

whom there was no clear explanation in the medical chart for the associated CFM code. Non-cases identified by the 744.23 code either had a syndrome involving microtia (e.g., Treacher Collins) or were individuals with anomalies of the ear that are not by definition considered microtia (e.g.: small ears) (Table 3)..

Discussion

Ascertainment of cases with congenital anomalies using ICD-9-CM codes is common in epidemiologic studies (Frohnert et al., 2005; Frohnert et al., 2005; Juhn et al., 2011; Quan et al., 2008; Rasmussen and Moore, 2001) and in birth defects surveillance systems reliant on passive case ascertainment methods, e.g., by linking hospital discharge records, vital statistics, or other administrative data (NBDPN, 2011; Rasmussen and Moore, 2001). Standardized methods for population-based ascertainment have not yet been widely used in CFM research. This is likely attributable to the lack of (1) diagnostic criteria for CFM and (2) ICD codes with high sensitivity and positive predictive values. The absence of a specific ICD-9-CM code for CFM could result in under- or overestimation of cases depending on the ICD-9-CM codes chosen for a given study.

Ideally, ascertainment of cases of CFM through healthcare databases and birth defects surveillance programs could be accomplished using one or more code combinations with high sensitivity and specificity. To help address this issue, modifications to the 1979 British Pediatric Association (BPA) Classification of Diseases and the World Health Organization's 1979 ICD-9-CM codes for birth defects were made by the Division of Birth Defects and Developmental Disabilities, National Center on Birth Defects and Developmental Disabilities, Centers for Disease Control and Prevention (CDC, 2007). These more specific codes are now employed by many active birth defects surveillance programs. Unfortunately, these codes are not widely used by healthcare facilities in the U.S.

In this study, the heterogeneity we observed in a systematic review of charts from two craniofacial centers suggests that ICD-9-CM codes, even in combination, do not appear to have sufficiently high sensitivity and positive predictive value to be used as the sole method of CFM case ascertainment for studies regarding CFM. Previous reports regarding the validity of ICD-9-CM codes for other birth defects, such as congenital heart disease, have also identified low sensitivity and a high false positive rate (Frohnert et al., 2005; Juhn et al., 2011; Quan et al., 2008; Rasmussen and Moore, 2001). Therefore, data ascertained through ICD-9-CM codes still require validation of clinical phenotypes through direct evaluation or medical record review by trained medical and health care professionals for studies when an accurate diagnosis is essential. This would not be a significant limitation if cases represented a high percentage of records flagged for review. However, this percentage was 12% or less at both centers, which suggests that the added requirement for a validation process may make this ascertainment method costly and inefficient.

Given the absence of a specific diagnostic code for CFM, the heterogeneity in coding practices we observed among medical centers is understandable, since centers or individual practitioners must subjectively decide which code fits best. These judgments appropriately differ depending on the patients' specific CFM-related features, such as for patients with and without microtia. In addition, while some practitioners will code every feature present in the patient, others will chose codes based on severity of the anomalies, or the indication for the current clinic visit. For example, a surgeon planning an ear reconstruction for a patient would likely chose the code for microtia, while another surgeon who operates on the same patient to repair a lateral oral cleft may opt to use the code for macrostomia.

A potential limitation of this study is that healthcare providers who care for CFM patients might assign ICD-9-CM codes other than those we evaluated. We relied on an expert panel of subspecialists who care for patients with CFM at craniofacial centers across the U.S. to determine the CFM eligibility criteria and identification of ICD-9-CM codes used for CFM. Our panel included clinicians from UNC and SCH to identify the most common codes they and their colleagues have used to denote CFM or its features. In addition, we thoroughly searched the ICD-9-CM for additional codes that could match the eligibility criteria. Thus, although use of other codes would mean some CFM cases could have been missed, it is unlikely that any other codes were used in a sufficiently high proportion to invalidate the study results. Indeed, such further variability supports the conclusion that a specific code is needed.

Including more centers in the study may have provided additional insight into the variability of coding by different centers. However, both craniofacial centers provide care for a large population of children with CFM and have an interdisciplinary model of care with multiple professionals assigning codes for patients. Therefore, we captured a wide range of codes used for CFM, and again, adding centers would only have increased the already high heterogeneity.

Making a diagnosis of CFM relying solely on the medical chart review can be challenging because the gold standard for any diagnosis is the medical examination. Because we did not perform physical examinations, it is possible that some CFM patients were misclassified as non-cases. This could have happened if, for example, a patient with facial asymmetry also had an unreported epibulbar dermoid. Although we cannot disregard such misclassification as a potential limitation, its impact on the study results was minimized by limiting the search to hospitals with craniofacial centers.

This is the first study, to the best of our knowledge, to address the use of ICD-9-CM codes for patients with CFM. A strength of the study was the development of a CFM case definition by consensus among a large, diverse panel of experts. In addition, charts were thoroughly reviewed by experienced providers by using a formally designed and structured medical chart abstraction form. Both the CFM case definition and the structured abstraction form are used in the FACIAL network studies and could potentially be used by other research teams interested in CFM. We would share the chart abstraction form and other data collection protocols to further CFM research efforts in other groups.

The study results suggest that ICD-9-CM code-based ascertainment of CFM might lead to under- or overestimation of the prevalence of this condition, depending on which codes are used. These results clearly indicate the need for a specific CFM code to facilitate CFM surveillance and research. The ICD-10 system has already been adopted in several countries, and the U.S. will incorporate this system into clinical billing systems in the near future. Though the ICD-10 system includes much more specific codes for many conditions, it has little impact on CFM coding. For instance, the new code Q87.0 “Congenital malformation syndromes predominantly affecting facial appearance” is more specific than the ICD-9-CM codes 756.0 and 754.0. It is recommended that this code be used for Goldenhar syndrome and perhaps could be used to denote CFM more generally. The code remains non-specific, however, and also includes unrelated conditions such as Moebius, oro-facial-digital, and CHARGE syndromes. Thus relying on this new code would still require medical records review to limit ascertainment specifically to CFM cases.

Even if a specific code for CFM were to be proposed immediately, it would not be implemented for at least several years. Findings from this study suggest codes that might be most efficient for surveillance and research on CFM (Table 4). Specifically, 744.01 (anotia)

and 744.23 (microtia) presented with reasonable sensitivity and positive predictive value and may be the most useful. Nevertheless, they cannot be the sole codes because many patients have no ear abnormalities. Codes such as 754.0, 756.0 and 744.1 showed better positive predictive value when used in combination and might be used for CFM patients in combination with at least one other code to improve efficiency of case ascertainment through ICD codes. The drawback of this approach is that only individuals assigned with two or more codes (41% of CFM patients in our sample) would be ascertained in this way. In summary, investigators should consider available resources, number of charts to be reviewed, and study objectives in deciding which codes to include in the search.

We found that relatively labor-intensive, active ascertainment techniques are currently needed to capture cases of CFM. Multiple codes are necessary because none of the ICD-9-CM codes alone ascertained all infants with CFM. In addition, the high number of non-cases resulting from a search based on the current ICD-9-CM codes demonstrated the need for physician confirmation through medical records review. As there are limited resources available for medical research and birth defects surveillance of CFM, such additional efforts will often not be possible. Lack of specific ICD-9-CM codes for CFM will continue to impede surveillance and research on this condition. The condition's prevalence, clinical relevance, and need for complex long-term healthcare warrant the development of a CFM-specific diagnostic code.

Supplementary Material

Refer to Web version on PubMed Central for supplementary material.

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Table 1

Facial Asymmetry Collaborative for Interdisciplinary Assessment and Learning (FACIAL) craniofacial microsomia: features included in case eligibility criteria and the corresponding ICD-9-CM codes.

Inclusion Criteria*‡	ICD-9-CM Code	ICD-9-CM Description
Epibulbar dermoid	743.8	Other specified congenital anomalies (CAs)* of eye
Microtia/Anotia	744.0	CAs of ear causing impairment of hearing
	744.01	Congenital absence external ear
	744.02	Other CAs of external ear with impairment of hearing
	744.09	Other CAs of ear causing impairment of hearing
	744.21	Congenital absence of ear lobe
	744.23	Microtia
Preauricular tag	744.1	Accessory auricle
Macrostomia	744.83	Macrostomia
Facial asymmetry	754.0	Musculoskeletal deformities of skull face and jaw
	756.0	CAs of skull and face bones
Facial tag	--------	---------

Table 2

Frequencies, sensitivity and positive predictive value (PPV) of the selected ICD-9-CM codes and numbers of codes per individual identified through hospital discharge record queries at the University of North Carolina and Seattle Children's Hospital among patients identified as possibly having craniofacial microsomia (CFM).

	University of North Carolina				Seattle Children's Hospital			
	Individuals with any of the selected ICD-9-CM codes*		Sensitivity	PPV	Individuals with any of the selected ICD-9-CM codes*		Sensitivity	PPV
	CFM (N=93)	no-CFM (N=706)			CFM (N=32)	no-CFM (N=468)		
ICD-9-CM codes**	**n (%)**	**n (%)**	**% (95% CI)**	**% (95% CI)**	**n (%)**	**n (%)**	**% (95% CI)**	**% (95% CI)**
744.01	--	--	--	--	6 (19)	1 (0)	19 (7–36)	86 (42–100)
744.02	9 (10)	1 (0)	10 (5–18)	90 (56–100)	9 (28)	8 (2)	28 (14–47)	53 (28–77)
744.09	1 (1)	1 (0)	1 (0–6)	50 (1–99)	--	--	--	--
744.1	--	--	--	--	2 (6)	15 (3)	6 (1–21)	12 (2–36)
744.23	76 (82)	6 (1)	82 (72–89)	93 (85–97)	23 (72)	5 (1)	72 (86–53)	82 (63–94)
744.83	5 (5)	1 (0)	5 (2–12)	83 (36–100)	0 (0)	2 (0)	--	--
754.0	33 (36)	664 (94)	36 (26–46)	5 (3–7)	1 (3)	254 (54)	3 (0–16)	<1 (0–2)
756.0	17 (18)	48 (7)	18 (11–28)	26 (16–39)	12 (38)	209 (45)	38 (21–56)	5 (3–9)
Combination of codes**								
744.01 & 744.02	0 (0)	n.a.	--	--	2 (6)	n.a.	6 (1–21)	67 (9–99)
744.01 & 744.23	0 (0)	n.a.	--	--	5 (16)	n.a.	16 (5–33)	100 (48–100)
744.01 & 756.0	0 (0)	n.a.	--	--	3 (9)	n.a.	9 (2–25)	75 (19–99)
744.02 & 744.1	0 (0)	n.a.	--	--	1 (3)	n.a.	3 (0–16)	100 (3–100)
744.02 & 744.23	6 (7)	n.a.	7 (2–14)	86 (42–100)	9 (28)	n.a.	28 (14–47)	90 (56–100)
744.02 & 744.83	1 (1)	n.a.	1 (0–6)	100 (3–100)	0 (0)	n.a.	--	--
744.02 & 754.0	4 (4)	n.a.	4 (1–11)	100 (40–100)	0 (0)	n.a.	--	--
744.02 & 756.0	4 (4)	n.a.	4 (1–11)	100 (40–100)	3 (9)	n.a.	9 (2–25)	75 (19–99)
744.1 & 744.23	0 (0)	n.a.	--	--	2 (6)	n.a.	6 (1–21)	67 (9–99)
744.1 & 756.0	0 (0)	n.a.	--	--	1 (3)	n.a.	3 (0–16)	50 (1–99)
744.23 & 744.83	3 (3)	n.a.	3 (1–9)	100 (29–100)	0 (0)	n.a.	--	--
744.23 & 754.0	21 (23)	n.a.	23 (15–32)	96 (77–100)	0 (0)	n.a.	--	--
744.23 & 756.0	10 (11)	n.a.	11 (5–19)	77 (46–95)	5 (16)	n.a.	16 (5–33)	83 (36–100)

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	University of North Carolina				Seattle Children's Hospital			
	Individuals with any of the selected ICD-9-CM codes*		Sensitivity	PPV	Individuals with any of the selected ICD-9-CM codes*		Sensitivity	PPV
	CFM (N=93)	no-CFM (N=706)			CFM (N=32)	no-CFM (N=468)		
744.83 & 754.0	2 (2)	n.a.	2 (0–8)	100 (16–100)	0 (0)	n.a.	--	--
744.83 & 756.0	2 (2)	n.a.	2 (0–8)	100 (16–100)	0 (0)	n.a.	--	--
754.0 & 756.0	8 (9)	n.a.	9 (4–16)	47 (23–72)	0 (0)	n.a.	--	--
Number of codes per individual								
1	55 (59)	692 (98)	n.a.	n.a.	19 (59)	444 (95)	n.a.	n.a.
2	30 (32)	13 (2)	n.a.	n.a.	7 (22)	22 (5)	n.a.	n.a.
3	7 (8)	1 (0)	n.a.	n.a.	4 (13)	2 (0)	n.a.	n.a.
>=4	1 (1)	--	n.a.	n.a.	2 (6)	0 (0)	n.a.	n.a.

n.a.: not applicable

CI: Confidence Interval

*There were no participants assigned with the ICD-9-CM codes: 743.8 744.0, and 744.21 either at UNC or SCH.

**Numbers do not add up to the totals because all possible two CFM-codes are shown, ie, if an individual had 3 codes assigned there are three possible pairwise combinations, so that individual is listed three times.

Table 3

Diagnoses of individuals that did not meet Facial Asymmetry Collaborative for Interdisciplinary Assessment and Learning (FACIAL) craniofacial microsomia eligibility criteria

	University of North Carolina		Seattle Children's Hospital	
Diagnosis	**N (%)**	**ICD-9-CM Codes**	**N (%)**	**ICD-9-CM Codes**
Skull Deformity	493 (70)	744.83, 754.0, 756,0	213 (46)	754.0, 756.0
Dysmorphic features	6 (1)	754.0	46 (10)	744.1, 754.0, 756.0
Macrocephaly	4 (1)	754.0	49 (11)	754.0, 756.0
Known syndrome	33 (5)	744.02, 744.09, 744.23, 754.0, 756.0	39 (8)	744.02, 744.1, 744.23, 744.83, 754.0, 756.0
Craniosynostosis	54(7.6)	754.0, 756.0	30 (6)	754.0, 756.0
Ear anomaly, not microtia	--	--	8 (2)	744.01, 744.02, 744.1, 744.23, 756.0
Cleft lip and palate	15 (2)	754.0	9 (2)	754.0, 756.0
Cleft palate	--	--	3 (1)	754.0, 756.0
Robin sequence	--	--	9 (2)	754.0, 756.0
Facial asymmetry	28 (4)	754.0	4 (1)	754.0, 756.0
Preauricular tag	1 (0)	754.0	7 (2)	744.1
Preauricular pit	1 (0)	754.0	--	--
Epibulbar dermoid	1 (0)	754.0	--	--
Eye anomaly	6 (1)	754.0	--	--
Hearing loss	2 (0)	754.0	3 (1)	744.02, 754.0
Trauma	3 (0)	754.0	3 (1)	754.0, 756.0
Other	32 (5)	754.0	25 (5)	744.02, 744.1, 754.0, 756.0
Unknown	25 (4)	754.0	--	--
Miscode	14 (2)	754.0	20 (4)	744.1, 754.0, 756.0

Multiple codes are listed for some diagnoses because for these diagnoses, providers used one of several different ICD-9-CM codes; this is due to the lack of specificity of ICD-9-CM codes for these conditions.

Table 4

Utility of ICD-9-CM codes for identifying craniofacial microsomia (CFM) for surveillance and research programs

ICD-9-CM code	Proposed decisions for ascertainment of CFM cases	Rationale
743.8	No	None met eligibility criteria
744.01	Yes	Reasonable[≠] sensitivity and positive predictive value at SCH
744.02	Yes	Reasonable[≠] sensitivity and positive predictive value at SCH and UNC The combination with another CFM code [*] improves PPV [**]
744.09	No	Only one individual assigned with this code met eligibility criteria
744.23	Yes	High sensitivity and specificity
744.1	Yes, in combination with another CFM code [*]	The combination with another CFM code [*] results in reasonable PPV at SCH
744.83	Yes	High PPV at UNC
754.0	Yes	Reasonable sensitivity at UNC The combination with another CFM code [*] improves PPV
756.0	Yes	Reasonable[≠] sensitivity and PPV The combination with another CFM code [*] improves PPV

[*] For example: 744.23 or 756.0

[**] PPV = positive predictive value

[≠] Reasonable is defined as a value of >15% in at least one institution