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Delayed diagnosis and underreporting of congenital anomalies associated with oral clefts in the Netherlands: A national validation study☆

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Summary *Objective:* Since 1997, the 15 Dutch cleft palate teams have reported their patients with oral clefts to the national oral cleft registry (NVSCA). During the first visit of the patient to the team — which is usually within the first year of life — the oral cleft and associated congenital anomalies are recorded through a unique recording form by a plastic surgeon/orthodontist/paediatrician. In this study, we evaluated the quality of data on congenital anomalies associated with clefts.
Methods: We drew a random sample of 250 cases registered in the national database with oral clefts from 1997 through 2003; of these, 13 were excluded. Using two independent reregisters derived from two-phased medical data review, we analysed whether associated anomalies were correctly diagnosed and recorded.
Results: The agreement on associated anomalies between the NVSCA and medical data ranged from moderate to poor (kappa 0.59 to 0). Seventy-seven percent of the craniofacial anomalies were underreported in the NVSCA: 30% due to delayed diagnosis and 47% due to deficient recording. Additionally, 80% of the associated anomalies of other organ systems were underreported: 52% due to delayed diagnosis and 28% due to deficient recording. The reporting of final

☆ This work has not been presented at any congress yet.

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diagnoses was somewhat better; however, 54% were still underreported (24% delayed diagnosis and 30% deficient recording). The rate of overreporting was 1.6% or lower.
Conclusion: Congenital anomalies associated with clefts are underreported in the NVSCA because they are under diagnosed and deficiently recorded during the first consultations with the cleft palate teams. Our results emphasise the need for routine and thorough examination of patients with clefts. Team members should be more focussed on co-occurring anomalies, and early genetic counselling seems warranted in most cases. Additionally, our findings underline the need for postnatal follow-up and ongoing registration of associated anomalies; reregistration in the NVSCA at a later age is recommended.

Oral clefts — one of the most common birth defects in humans — range from mild types to complete clefts affecting the lip/alveolus/palate. Although many genetic and environmental factors (e.g., smoking and nutrition) have been found to contribute to their development,[1,2] the aetiopathogenesis of oral clefts is still poorly understood. Oral clefts are frequently associated with other congenital anomalies, often as part of a syndrome or chromosomal defect. However, the proportion of individuals with additional anomalies varies greatly between studies (3—63%) and appears to be related to time of registration and how data have been collected.[3] It has also not conclusively been established whether oral clefts are related to specific types of associated anomalies and there are differences among reports concerning which organ systems are most commonly affected.[4,5] Given that children with oral clefts associated with other congenital anomalies have much higher morbidity and mortality throughout life than do individuals with isolated clefts,[6–8] early and sound diagnosis of co-occurring anomalies is of paramount importance. Furthermore, complete and accurate data on oral clefts and their associated anomalies are needed to facilitate further genetic and aetiopathological studies and prevention of clefts.[1]

Since 1997, the 15 multidisciplinary cleft palate teams in the Netherlands have reported their new pre-surgery patients with clefts to the national oral cleft registry, which is maintained by the Dutch Association for Cleft Palate and Craniofacial Anomalies (NVSCA). These teams treat virtually all surviving children with clefts who reside in the Netherlands. Using a unique detailed recording form based on the embryology of the head and neck area, oral clefts and their associated congenital anomalies are recorded.[9,10] Depending on the team, the form is completed by a plastic surgeon (nine teams), orthodontist (three teams) or paediatrician (three teams) during the first visit of the patient to the team, which is usually within the first year of life.

As the main purposes of the NVSCA are to optimise diagnostics, treatment and prevention of oral clefts and to provide a solid basis for clinical, epidemiological and fundamental research, it is crucial that data provided by this registry are of high quality. Early and sound diagnosis and complete reporting of oral clefts and their associated anomalies are essential to maintain high standards of cleft care and data quality. Previously, it was shown that oral clefts — especially those types that are readily diagnosed at birth — are recorded completely and accurately in the NVSCA.[10–12] It is not known, however, whether associated anomalies are also correctly recorded by the 15 cleft palate teams. Because not all associated anomalies are detectable at birth or in the neonatal period,[3,13] these anomalies might be underreported due to delayed diagnosis. Another factor that might cause underreporting of associated anomalies is deficient (incomplete/incorrect) recording by the consulting physicians.[14]

In this article, the last of three articles validating the NVSCA registry,[10,11] we evaluated the quality of data on congenital anomalies associated with clefts by validating I) additional anomalies of the head and neck area, II) additional anomalies of other organ systems and III) final diagnoses (including syndromes and chromosomal defects). Using two independent reregisters derived from two-phased medical data review, we investigated whether these anomalies were diagnosed and recorded correctly during the first consultations with the Dutch cleft palate teams.

Material and methods

NVSCA registry

The methodology by which the NVSCA registry was established is described in detail elsewhere[9–11] and is summarised here. The NVSCA is an anonymous registry. All Dutch cleft palate teams report their new patients — before they have an oral cleft operation — through a standard NVSCA form; a manual is available.[9,10] The recording form is composed of three parts. The first is a general section for infant/parental characteristics. The second part consists of a two-dimensional table based on the embryology of the head and neck area, in which oral clefts and any associated craniofacial anomaly (e.g., mandibular hypoplasia or congenital ear anomalies) can be recorded in detail. As shown in Figure 1, the *x*-axis depicts the topographic-anatomical structures, the *y*-axis the morphologic features and the checking boxes represent the side. The last (third) part gives space for verbatim descriptions of both major and minor congenital anomalies of other parts of the body and final diagnoses, including syndromes and chromosomal defects.

Note that the NVSCA does not have active follow-up of patients and that no data from other sources are included.