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Proposal and practicalities for a global registry and database on craniofacial anomalies

7.1 A global registry for craniofacial anomalies

The basic idea, as discussed at the Baurú meeting, is to create a master database – or registry of registries - that maintains the individuality and independence of each of the contributing programmes with all their valuable diversities and flexibilities, and is capable of being adapted to cover the large variety of different social, economic, political and cultural situations. In spite of the diversities, a common core of similar standard data on clefting and other craniofacial birth defects can be found and successfully shared in a central pool for the purpose of global research.

7.1.1 *Rationale and aims*

The rationale for the registry is that it will identify global variability in the prevalence of craniofacial birth defects, estimate the burden of need for public health services, identify priorities and underpin research initiatives that will address primary and tertiary prevention.

This proposal can be envisaged as a disease-specific “registry of registries”, or a global coordinating registry, dealing with craniofacial congenital anomalies in general, but starting specifically with the most frequent and relevant anomalies: OC, including CL/P, and CP. Other CFA could be added in the future.

BOX 18

Aims and objectives of a CFA registry

The general objective of a global registry for CFA is to systematically “register” CFA cases within the classical definition of a register, namely, to detect, enlist and follow up (Last, 1995), in order to build a worldwide collaborating network and an active database, capable of providing material and coordinating the following activities:

- **Surveillance:** Monitoring, searching for geographical clusters, etc.
- **Promotion:** Creating, helping and supporting local registers and centres.
- **Expertise:** Providing expertise from a worldwide net of experts and an available task force.
- **Prevention:** Creating, recommending and conducting public health activities.
- **Community outreach:** Actively interacting with support organizations.
- **Education and training** with worldwide coverage.
- **Research:** Assisting with research, such as temporary activities aimed at specific objectives.

7.1.2 Structure

Based on the principle that no ideal registry exists, the main characteristic should be *flexibility* in order to benefit from the existing diversity of methodologies of registration, allowing for better adaptation to the range of economic development worldwide, different cultures, and levels of interest in this type of disease (Källén et al., 1992).

The global registry could enrol participating members into different levels of activities, with the understanding that a given participant may be suitable for only particular activities. Several levels of participation could be established.

BOX 19**Possible levels of participation**

- **Surveillance, including periodic (quarterly and/or annual) exchange of data for monitoring purposes and for other surveillance activities:** Population-based and governmental systems would be more appropriate for this activity. A minimal core of exchanged data elements would be provided (*see para. 7.1.3 below*).
- **Registry and follow-up of cases, also specialized clinics with no denominators (e.g., Centrinho in Baurú):** Hospital-based systems, isolated clinics and other case repositories would be better adapted to this activity.
- **Research, including different levels of research (epidemiology, clinical dysmorphology, family studies, molecular analysis, etc.):** The feasibility of participating in this activity would depend on the specific project design, and for any particular research, the contribution of only some member systems would be expected.
- **Other activities, such as public health, community outreach, education and training, etc.,** could allow for the participation of all members, and membership could even extend to parent-patient associations, and other organizations dealing with CFA.

7.1.3 Core data elements

An agreed core or minimum dataset is an essential resource for surveillance/registration of a birth defect. This data must be available as a central and accessible resource, along with recommendations that may assist in those areas of the world that do not have such data available (*see Annex*).

BOX 20**Core data elements for CFA cases, case by case**

- identity number,
- birth date (month and year),
- place of birth (municipality),
- place of residence of mother during pregnancy (municipality),
- status (live birth, stillbirth or interrupted pregnancy),
- sex (male, female, intersex),
- twinning (no, yes/like-sexed, yes/unlike-sexed, yes/unknown sex),
- birth weight (grams),
- mother's age (years),
- parity (gravidity or birth order).
- **For all births: aggregated monthly data (denominators).**

For monitoring purposes, the same information should be available for all examined births. A summary table recording the data collected could be used by each registry on a monthly basis, even if it is only submitted annually (see Annex, Figure 6).

7.1.4 A possible corporation

A Global Registry of Craniofacial Anomalies could be organized under the umbrella of the World Health Organization, using existing facilities. It could be housed in Rome by the ICBD (International Centre for Birth Defects, which is the ICBDMS headquarters) where it could profit from the following aspects of the ICBD:

- global coverage,
- methodological flexibility,
- WHO-NGO status and inclusion of several WHO Collaborating Centres,
- recognized international expertise since 1974.

To hasten implementation of this new registry, the following four major networks that detect birth defects could be invited to provide data as initial members: ECLAMC, EUROCAT, ICBDMS, NBDPN.

7.2 Proposal for the WHO/NIH Global Registry initiative

7.2.1 Create an international registry of oral clefts

Multiple sources will be used to collect cases:

- existing birth-defect registries,
- surgery clinics,
- parents' associations,
- any other possible source/s, to be identified country by country, area by area.

For each case a minimal data set of information will be collected: patient's identity (ID) and basic personal characteristics, type of cleft, possible associated anomalies. Where possible, or for selected sources only, more complete information will be collected, including family history, postnatal care and, perhaps, blood spots. Basic data will be used for simple descriptive epidemiology, but more sophisticated studies might be possible using more detailed data from a subset of participating registries.

A worldwide registry of oral clefts could almost be organized under the umbrella of WHO, through the use of existing facilities of the ICBDMS. This registry could also be based in Rome, Italy at the ICBD which is currently the ICBDMS headquarters.

The ICBD has the considerable advantages of:

- global coverage ,
- methodological flexibility,
- WHO-NGO status and inclusion of several WHO Collaborating Centres,
- recognized international expertise since 1974.

In the first instance, four major existing networks for detecting birth defects could be invited to provide data to the Global Registry at ICBD as initial members for promptness of implementation, namely: ECLAMC, EUROCAT, ICBDMS, NBDPN.

7.2.2 Create and maintain a directory of resources

A directory of resources would include a database on all possible information and resources on oral clefts around the world (published articles, books and other relevant documents; lists of institutions and researchers; funding bodies; treatment centres and other bodies concerned with CFA research). The following areas would be considered:

- **Genetics:** An update on research programmes, research teams and genetic clinics that at present aim to identify etiologic causes of non-syndromic OC through family-based studies and search for candidate genes.
- **Prevention:** Special attention would be given to reviewing evidence regarding the role of specific maternal nutritional factors and to making recommendations on the resources needed to implement international collaborative studies of cleft prevention with common core protocols.
- **Gene-environment interactions:** Descriptions of the state-of-the-science, would describe and highlight relevant research.
- **Treatment:** Identification of optimal clinical interventions for the management of oral clefts (evidence-based care), identification and dissemination of strategies to optimize the quality of care delivered, identification of strategies to increase the availability of care to all affected citizens in the world.

7.2.3 International Database on Craniofacial Anomalies

The International Database of Craniofacial Anomalies (IDCFA) is an initiative to help researchers all over the world to better understand the epidemiology of CFA and the epidemiology of health care related to persons with a CFA. It is a database with data being contributed by any collaborating organization in the world. Representatives of all the major

organizations contributing to the database serve on its Steering Committee: ECLAMC, EUROCAT, ICBD, ICBDMs, NBDPN, and WHO's Human Genetics Programme (WHO-HGN).

ICBD is the coordinating office of the IDCFA (Coordinators: Pierpaolo Mastroiacovo and Elisabeth Robert-Gnansia), and most of the members of the above-mentioned member-programmes are contributing registries. Data are stored in an agreed-upon format, and the database is updated case by case every sixth month. The database is accessible through a password to collaborators who wish to consult or use it for a special study, after permission is obtained from the Steering Committee. Confidentiality and data protection are guaranteed by a complete set of data security rules, established for the use of any data in the frame of ICBD.