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A challenge for comparing international studies

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COMMENTARY

Lack of consensus on the definition of severe congenital heart defects for research: A challenge for comparing international studies

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In this issue of *Paediatric and Perinatal Epidemiology*, Gimeno and colleagues[1] examine trends in the survival of children born with severe congenital heart defects (CHD) by gestational age in all of England using administrative hospital data. The main strengths of the study are the use of a national healthcare database that allows for the inclusion of a much larger population in the study than in traditional research using medical records as the primary data source and the almost complete follow-up of the children born with severe CHD. The main limitation is the exclusion of 19.9% of the births due to missing or implausible gestational age, which led to the exclusion of 23.2% of the children with a diagnosis of severe CHD within the study period.

Differences in the prevalence of CHD across regions and countries are mainly explained by variations in the inclusion of small ventricular septal defects (VSDs) and access to and waiting time for echocardiography.[2,3] Studies may focus on only severe CHD, which may be ascertained more comparably. However, there is no international consensus on the definition of severe CHD. The study by Gimeno et al. defined severe CHD according to the 2014 definition of the European network of population-based registries for the Epidemiological Surveillance of Congenital Anomalies (EUROCAT)[4,5] and further included all children that had cardiac surgery or intervention within the first 5 years for less severe CHD such as septal defects. In fetal medicine, the definition of major CHD includes all cardiac defects that are lethal or require surgery or cardiac intervention within the first year after birth, and the definition is used when estimating the prenatal detection rates of CHD.[6] This definition requires access to follow-up data on survival and surgery within the first year. The population-based study by Gimeno et al.[1] included children with surgery or cardiac intervention performed up to 5 years of age which means that more children were included than if the definitions from fetal medicine were applied.

The first EUROCAT definition of severe CHD was based on the study on prevalence and perinatal mortality for CHD in Europe from 2000 to 2005.[3] After grouping the study cases with a specific ICD code for CHD into three groups based on perinatal mortality, it was clear that all cardiac defects in the most severe group were univentricular heart defects. The group with intermediate perinatal mortality included cardiac defects that could be corrected by surgery to normal anatomy and the less severe group included atrial septal defects (ASD), VSD, and pulmonary valve stenosis. After the study, the EUROCAT Coding and Classification Committee defined the severe CHD subgroup for prevalence and surveillance as the ICD codes for the cardiac anomalies included in the two groups with the highest perinatal mortality. The severe CHD subgroup was revised in 2014 with additional ICD codes for more rare cardiac anomalies with specific ICD codes such as aortic arch anomalies. A recent review of EUROCAT cases for the severe CHD subgroup showed that the cases included with mitral valve insufficiency often had secondary valve insufficiency due to dilatation of the left ventricle. Therefore, in the most recent version of the EUROCAT subgroups published on the EUROCAT website in 2022, the severe CHD subgroup does not include the code for mitral valve insufficiency.[4]

The advantage of using the EUROCAT definition of severe CHD in research based on healthcare databases is that the classification is based on the ICD codes only. The disadvantage is that healthcare databases may contain inaccurately recorded ICD codes and the coding of CHD is difficult and challenging for noncardiologists. Studies have shown that for 90% of the children with a code for CHD in hospital databases the CHD diagnosis was correct.[7–9] As children

have ICD codes for every hospital contact, the codes may not be the same from the initial suspected diagnosis around the birth to the follow-up after surgery and some codes may be more unspecified such as Q248 (other specified CHD) and Q249 (unspecified CHD). The study by Gimeno et al.[1] included all children with any ICD code up to age 5 years for the specific severe CHD included in the EUROCAT definition. This may result in the inclusion of some children where the diagnosis of severe CHD was a data entry error or later was corrected to a less severe CHD. Other options may be to use the CHD diagnosis given by paediatric cardiologists as the diagnosis for inclusion or set as a requirement that the specific CHD diagnosis is present in more than one hospital contact. A study from Australia showed that by restricting the inclusion of children with multiple diagnoses in separate admissions reduced the over-recording in the hospital databases from 10% to 2% for children with any CHD.[6] Researchers using healthcare databases to study children with severe CHD should be aware of the important step of developing the inclusion criteria based on the optimal use of the data available. Frequency lists with ICD codes for each child will be valuable to know the proportion of children with multiple codes for severe CHD and the proportion of children with the codes for unspecified and other specified CHD only before defining the inclusion criteria and the analysis for the study.

The lack of an international consensus on the definition of severe CHD for research limits the comparisons across studies and populations. The prevalence of severe CHD in the study by Gimeno et al. was 3.6 per 1000 live births compared to the EUROCAT prevalence of severe CHD at 1.9 per 1000 live births for the same birth years.[4] This indicates that around half of the children in the study by Gimeno et al. had septal defects only with an expected high survival rate.

For the identification of children with severe CHD in the study by Gimeno et al., 47% had both a diagnostic code (ICD10) and a procedural code, 30% had procedural codes only and 23% had diagnostic codes only.[1] Among the children with procedural codes only, a high proportion is expected to have septal defects that are not included in the EUROCAT definition of severe CHD, but there may also be children with severe CHD that are not coded correctly in the hospital databases or have the codes only for unspecified or other specified CHD. The 23% of children included with a diagnostic code only will include children dying before surgery but may also include children with less severe CHD not requiring intervention but have one or more codes for a severe CHD in the hospital databases. Some of them may have the code for mitral valve insufficiency as the study used the older EUROCAT definition of severe CHD.

The citizens in Europe seem to be positive about the use of healthcare data for research as long as it is for the common good.[10] Many more studies will be published with outcome data for children with severe CHD based on big data from healthcare databases. Classifying severe CHD in healthcare databases based on more variables than the ICD codes is complicated and for every additional variable added, there will be the issue of incomplete data. It would be good if future studies used the same definition of severe CHD to make the outcome for these children comparable across countries. The EUROCAT definition of severe CHD is simple to use as it is based on ICD codes only and is publicly available at the EUROCAT website.[4]

ABOUT THE AUTHOR

Ester Garne is a Consultant Paediatrician at Hospital Lillebaelt and associate professor at the University of Southern Denmark. She is the registry leader of the EUROCAT registry for Funen and has been the Chair of the EUROCAT Coding and Classification Committee for 20 years. She has research collaboration with the international networks on cerebral palsy (SCPE and the Australian Cerebral Palsy register) and was a partner in the EU FP7 project EUROmediCAT 2011-2014. She is the clinical coordinator of the EUROlinkCAT study. The main research interests are congenital anomalies and perinatal epidemiology.

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CONFLICT OF INTEREST STATEMENT

The author declares no conflicts of interest.

DATA AVAILABILITY STATEMENT

No new data presented to share, only data from references.

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