British Isles Network of Congenital Anomaly Registers

BINOCAR Standard Operating Procedure for Data Quality Indicators

Instructions for the Registration and Surveillance of Congenital Anomalies in England and Wales

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1.1	30 January2015	First version put together by AS, AT and the BINOCAR Management Committee

Contents

Introduction

Complex factors influence the quality of data collected by a congenital anomaly register. How, where, and when diagnoses of congenital anomaly are made to residents of the region? How does the register obtain and verify this information and how is it coded? Register data are never a "perfect" description of which babies have congenital anomalies in the population and precisely what those anomalies are. Moreover, diagnostic definitions and methods change over time.

Registers may need to make difficult decisions about which types of information to prioritise in order to allocate limited resources, particularly whether to devote resources to collection of exposure information as well as diagnostic information. Some types of information are easier to obtain in some regions than in others, depending on specialist referral systems for affected children, confidentiality restrictions, what is recorded in medical notes, availability of computer databases, and willingness to collaborate of key professionals.

BINOCAR's policy is to strive for high quality, accompanied by transparency as to strengths and weaknesses in data quality. Running the analysis on these Data Quality Indicators (DQIs) and sharing them with the registers will allow them to evaluate their performance in relation to other registers, help focus the attention of registers on areas needing improvement, and will allow appropriate interpretations to be made of the results of the analyses. The DQIs only have relevance in relation to the objectives of BINOCAR. Different data strengths are needed to participate in timely statistical monitoring in relation to environmental teratogenic exposures, or to evaluate the population sensitivity of prenatal screening for detecting affected pregnancies, or to establish the prevalence of rare genetic syndromes.

The following areas are covered in this set of Data Quality Indicators on register data transmitted to EUROCAT:

- Completeness of case ascertainment
- Precision of diagnosis
- Completeness of information

The analysis for the DQIs will be run biannually after receipt of the updated data from EUROCAT on five years of data (by year of delivery/pregnancy end). Cases will be assigned to a register using the mother's postcode of residence at delivery/pregnancy end. Poisson regression will be used to identify any registers that have results statistically significantly higher or lower than the average for all registers.

The results of these analyses will be passed onto the registers for them to have a look at and give them the opportunity to comment on any indicators where they have been highlighted as statistically significantly different.

Annually at the AGM the results of this will be presented and will give the registers the opportunity to discuss with each other ways to improve the ascertainment and quality of the data.

Indicators

The EUROCAT list of DQIs was taken as a starting point, and their appropriateness and reason for being included were discussed for each DQI. Some of the EUROCAT DQIs were kept, some were adapted and some new ones were created. The following are the BINOCAR DQIs:

1 ASCERTAINMENT

1.1 Total congenital anomaly prevalence

Source of DQI:	EUROCAT
Rationale:	To look at the overall prevalence of congenital anomalies
Definitions:	All anomalies = Q-chapter, D215, D821, D1810, P350, P351, P371 and excluding minor anomalies listed in Appendix A. Cases = Live births (LB), stillbirths (24+ weeks' gestation, SB), late miscarriages (20-23 weeks' gestation, Misc) and terminations of pregnancy for fetal anomaly (all gestations, TOPFA).
Calculation:	Number of cases with an anomaly divided by the number of total births (LB and SB) in the whole population. Given per 10,000 total births.

1.2 Prevalence of malformed fetal death ≥ 22 weeks' gestation

Source of DQI:	modified EUROCAT
Rationale:	To look at prevalence that can be validated against another data source.
Definitions:	All anomalies = Q-chapter, D215, D821, D1810, P350, P351, P371 and excluding minor anomalies listed in Appendix A. Fetal deaths = SB and Misc
Calculation:	Number of fetal deaths (22+ weeks' gestation) and TOPFA (22+ weeks' gestation) with an anomaly divided by the number of total births in the whole population. Given per 10,000 total births. Compare to MBRRACE reference data.

1.3 Prevalence of Down syndrome adjusted for maternal age (at delivery)

Source of DQI:	EUROCAT
Rationale:	To look at the prevalence of a chromosomal anomaly that is not known to vary by region (once the main confounder has been corrected for)
Definitions:	Down syndrome = Q90 Cases = LB, SB, Misc & TOPFA Maternal age groups = <20, 20-24, 25-29, 30-34, 35-39, 40+
Calculation:	Observed number of cases with Down syndrome within each maternal age group divided by the expected number of cases (within the same time period) with Down syndrome within each maternal age group given that the prevalence for that register is the same as the overall prevalence. Given as a percentage. Expected number of cases will be calculated using the prevalence within each maternal age group for all registers combined and applied to the population of the register.

1.4 Prevalence of exomphalos - non chromosomal

Source of DQI: BINOCAR
Rationale: To look at the prevalence of a structural anomaly that is not known to vary by region
Definitions: Exomphalos = Q792
Cases = LB, SB, Misc and TOPFA
Non chromosomal = Case does not have a chromosomal anomaly. It includes isolated and cases with multiple anomalies
Calculation: Number of cases with isolated exomphalos divided by the number of total births in the whole population. Given per 10,000 total births.

1.5 Prevalence of congenital diaphragmatic hernia (CDH)

Source of DQI: BINOCAR
Rationale: To look at the prevalence of a structural anomaly that is not known to vary by region
Definitions: CDH = Q790 (isolated, multiple and chromosomal combined)
Cases = LB, SB, Misc and TOPFA
Calculation: Number of cases with CDH divided by the number of total births in the whole population. Given per 10,000 total births.

1.6 Prevalence of cleft palate (without cleft lip)

Source of DQI: modified EUROCAT
Rationale: To look at the prevalence of a postnatally diagnosed congenital anomaly
Definitions: Cleft palate = Q35
Cases = LB, SB, Misc and TOPFA
Calculation: Number of cases with cleft palate divided by the number of total births in the whole population. Given per 10,000 total births.

2 PRECISION OF DIAGNOSIS

2.1 Proportion of cases with multiple anomalies

Source of DQI: EUROCAT
Rationale: To look at individual register coding approaches
Definitions: All anomalies = Q-chapter, D215, D821, D1810, P350, P351, P371 and excluding minor anomalies listed in Appendix A.
Cases = LB, SB, Misc and TOPFA
Multiple = Two or more unrelated structural anomalies
Calculation: Number of cases with multiple anomalies divided by the number of cases with an anomaly. Given as a percentage.

2.2 Proportion of chromosomal cases (except T13, T18, T21) with a known recorded karyotype

Source of DQI: EUROCAT
Rationale: To make sure as much information as possible is provided on chromosomal anomalies
Definitions: Chromosomal cases (except T13, T18, T21) = Q92, Q93, Q96-Q99 and excluding Q936
Cases = LB, SB, Misc and TOPFA
Karyotype = Textual description of the karyotype from the cytogenetic laboratory (exact karyotypes or identified from PCR)
Calculation: Number of cases with a chromosomal anomaly (except T13, T18, T21) with known recorded karyotype divided by the number of cases with a chromosomal anomaly (except T13, T18, T21). Given as a percentage.

2.3 Proportion of selected 4-digit Q-BPA codes (Q3380, Q3690, Q3710, Q6140, Q6141)

Source of DQI:	modified EUROCAT
Rationale:	To make sure that specific anomalies are being coded correctly
Definitions:	Anomaly codes = Q33.80, Q36.90, Q37.10, Q61.40, Q61.41 Cases = LB, SB, Misc and TOPFA
Calculation:	Number of cases with specific anomaly codes divided by the total number of cases with the less specific related 3 digit Q code and the 4 digit Q code. Given as a percentage.

3 COMPLETENESS OF INFORMATION

3.1a-i Completeness of EUROCAT core variables

Variables:	sex, number of babies, number of malformed babies within multiple pregnancy, birth weight, gestation at birth, one week survival, timing of diagnosis, gestation of diagnosis if prenatal, maternal age (at delivery)
Source of DQI:	EUROCAT
Rationale:	To look at the completeness of the important and useful variables in the dataset
Definitions:	All anomalies = Q-chapter, D215, D821, D1810, P350, P351, P371 and excluding minor anomalies listed in Appendix A. Cases = LB, SB, Misc and TOPFA
Calculation:	Number of cases with an anomaly with a known value divided by the total number of cases with an anomaly (i.e. not known and missing values are excluded from the numerator).
Notes:	*Each variable to be presented separately.* *The denominator for the completeness of the number of malformed babies within multiple pregnancies variable will be only multiple pregnancies.* *The denominator for the completeness of the birth weight variable will be only live and still births.* *The denominator for the completeness of the gestation of diagnosis variable will be only prenatal diagnosed cases.* *The denominator for the completeness of the one week survival variable will be only live births.*

3.2a-c Completeness of BINOCAR core variables

Variables:	postcode at booking, postcode at delivery, ethnicity
Source of DQI:	BINOCAR
Rationale:	To look at the completeness of the important and useful variables in the dataset.
Definitions:	All anomalies = Q-chapter, D215, D821, D1810, P350, P351, P371 and excluding minor anomalies listed in Appendix A. Cases = LB, SB, Misc and TOPFA
Calculation:	Number of cases with an anomaly with a known value divided by the total number of cases with an anomaly (i.e. not known and missing values are excluded from the numerator).
Notes:	*Each variable to be presented separately.*

3.3a-cCompleteness of register core variables

Variables:	mother's NHS number, infant's NHS number, mother's date of birth
Source of DQI:	BINOCAR
Rationale:	To look at the completeness of the important and useful variables in the dataset.
Definitions:	All anomalies = Q-chapter, D215, D821, D1810, P350, P351, P371 and excluding minor anomalies listed in Appendix A. Cases = LB, SB, Misc and TOPFA
Calculation:	Number of cases with an anomaly with a known value divided by the total number of cases with an anomaly (i.e. not known and missing values are excluded from the numerator).
Notes:	*Each variable to be presented separately.* *The denominator for the completeness of the infant's NHS number variable will be only live births and stillbirths.*

3.4 Proportion of syndrome text complete

Source of DQI:	EUROCAT
Rationale:	To make sure as much information as possible is provided on anomalies, as it is important for coding cases and use in specific projects.
Definitions:	Cases = LB, SB, Misc and TOPFA Syndrome text = Variable that holds the textual description of the ICD10 code for the syndrome.
Calculation:	Number of cases with an ICD10 code for a syndrome with the text field complete divided by the total number of cases with an ICD10 code for a syndrome. Given as a percentage.

3.5 Proportion of malformation text complete

Source of DQI:	EUROCAT
Rationale:	To make sure as much information as possible is provided on anomalies, as it is important for coding cases and use in specific projects.
Definitions:	Cases =LB, SB, Misc and TOPFA Malformation text = Variable that holds the textual description of the first ICD10 code for an anomaly.
Calculation:	Number of cases with an ICD10 code for the first anomaly with the text field complete divided by the total number of cases with an ICD10 code for the first anomaly. Given as a percentage.

Appendix A – Minor anomalies list

The following table and footnotes are reproduced from EUROCAT Guide 1.4 Chapter 3.2.

	Specified ICD10-BPA – if present
Head	
Aberrant scalp hair patterning	
Flat occiput	
Dolichocephaly	Q67.2
Plagiocephaly – head asymmetry	Q67.3
Bony occipital spur	
Third fontanel	
Macrocephalus	Q75.3
Facial asymmetry	Q67.0
Compression facies	Q67.1
Other cong deformities of skull, face and jaw	Q67.4
Eyes	
Epicanthic folds	
Epicanthus inversus	
Upward slanting palpebral fissures	
Downward slanting palpebral fissures	
Short palpebral fissures	
Congenital ectropion	Q10.1
Congenital entropion	Q10.2
Other congenital malformation of eyelid	Q10.3
Dystopia canthorum	
Hypertelorism	Q75.2
Hypotelorism	
Stenosis of stricture of lacrimal duct	Q10.5
Synophrys	Q18.80
Blue sclera	Q13.5
Crocodile tears	Q07.82
Ears	
Primitive shape	Q17.3
Lack of helical fold	Q17.3
Asymmetric size	Q17.3
Posterior angulation	Q17.3
Microtia	Q17.2
Macrotia	Q17.1
Protuberant ears	Q17.3
Absent tragus	
Double lobule	Q17.0
Accessory auricle, preauricular appendage, tag or lobule	Q17.0
Auricular pit	
Preauricular sinus or cyst	Q18.1
Narrow external auditory meatus	

Low set ears	Q17.4
Bat ear, prominent ear	Q17.4
Unspecified and minor malformation of ear	Q17.9
Nose	
Small nares	
Notched alas	
Oral regions	
Borderline small mandible/ minor micrognathia	
Aberrant frenula	
Enamel hypoplasia	
Malformed teeth	
High arched palate	Q38.50
Tongue tie or cyst of tongue	Q38.1
Macroglossia	Q38.2
Macrostomia	Q18.4
Microstomia	Q18.5
Macrocheilia	Q18.6
Microcheilia	Q18.7
Ranula	
Neck	
Mild webbed neck	
Sinus, fistula or cyst of branchial cleft	Q18.0
Preauricular sinus or cyst	Q18.1
Other branchial cleft malformation	Q18.2
Congenital malformation of face and neck, unspecified	Q18.9
Torticollis	Q68.0
Hands	
Duplication of thumbnail	
Enlarged or hypertrophic nails	Q84.5
Single/abnormal palmar crease	Q82.80
Unusual dermatoglyphics	
Clinodactyly (5th finger)	
Short fingers (4.5th finger)	
Accessory carpal bones	Q74.00
Feet -Limb	
Syndactyly (2nd-3rd toes)	
Gap between toes (1st-2nd)	
Short great toe	
Recessed toes (4th, 5th)	
Enlarged or hypertrophic nails	Q84.5
Prominent calcaneus	
Clicking hip, subluxation of unstable hip	Q65.3-Q65.6
Metatarsus varus or metatarsus adductus	Q66.2
Hallux varus – other cong varus deformities of feet	Q66.3
Talipes or pes calcaneovalgus	Q66.4

Congenital pes planus	Q66.5
Metatarsus varus – other cong valgus deformities of feet	Q66.6
Pes cavus	Q66.7
Clubfoot of postural origin – other cong deformities of feet	Q66.8
Congenital deformity of feet, unspecified	Q66.9
Skin	
Haemangioma (other than face or neck)	
Pigmented naevus – cong non-neoplastic naevus	Q82.5
Naevus flammeus	Q82.50
Strawberry naevus	Q82.51
Lymphangioma	
Angioma	
Persistent lanugo	
Mongoloid spot (whites)	Q82.52
Depigmented spot	
Unusual placement of nipples	
Accessory nipples	Q83.3
Cafe-au-lait spot	
Skeletal	
Cubitus valgus	
Prominent sternum	Q67.7
Depressed sternum	Q67.6
Sternum bifidum	Q76.71
Shield like chest, other cong deformities of chest	Q67.8
Congenital deformity of spine	Q67.5
Genua valgum	
Genus varum	
Genu recurvatum	Q68.21
Congenital bowing of femur	Q68.3
Congenital bowing of fibula and tibia	Q68.4
Congenital bowing of long bones of leg, unspecified	Q68.5
Spina bifida occulta	Q76.0
Sacral dimple	
Cervical rib	Q76.5
Absence of rib	Q76.60
Accessory rib	Q76.62
Congenital lordosis, postural	Q76.43
Brain	
Arachnoid cyst	
Choroid plexus cyst	
Anomalies of septum pellucidum	
Cardiovascular	
Absence or hypoplasia of umbilical artery, single umbilical artery	Q27.0
Functional or unspecified cardiac murmur	
Patent ductus arteriosus if GA < 37 weeks	Q25.0 if GA <37 weeks
Peripheral pulmonary artery stenosis	

Patent or persistent foramen ovale	Q21.11
Pulmonary	
Accessory lobe of lung	Q33.1
Congenital laryngeal stridor	Q31.4
Laryngomalacia	Q31.4, Q31.5
Tracheomalacia	Q32.0
Azygos lobe of lung	Q33.10
Gastro-intestinal	
Hiatus hernia	Q40.1
Pyloric stenosis	Q40.0
Diastasis recti	
Umbilical hernia	
Inguinal hernia	
Meckel's diverticulum	Q43.0
Functional gastro-intestinal disorders	Q40.21, Q43.20, Q43.81, Q43.82
Transient choledochal cyst	
Anterior anus	
Renal	
Vesico-ureteral-renal reflux	Q62.7
Hydronephrosis with a pelvis dilatation less than 10 mm	
Hyperplastic and giant kidney	Q63.3
Single renal cyst	Q61.0
External genitals	
Deficient or hooded foreskin	
Undescended testicle	Q53
Unspecified ectopic testis	
Retractile testis	Q55.20
Hydrocele of testis	
Phimosis	
Bifid scrotum	Q55.21
Curvature of penis lateral	
Hypoplasia of penis	
Hymen imperforatum	Q52.3
Fusion of labia	Q52.5
Prominent labia minora	
Enlarged clitoris	
Vaginal skin tag	
Cysts of vulva	
Transient ovarian cyst	
Other	
Congenital malformation, unspecified	Q89.9
Chromosomal	
Balanced translocations or inversions in normal individuals	Q95.0, Q95.1

“Non-congenital” anomalies

Pyloric stenosis – there is controversy about the congenital nature of the majority of cases.
Patent ductus arteriosus in babies <37 weeks
Hydrocephaly where a result of preterm birth rather than congenital: all cases among preterm births should be thoroughly checked before registration.

Poorly specified anomalies

Functional or unspecified cardiac murmur
Laryngomalacia and tracheomalacia
Functional gastro-intestinal disorders
Undescended testicle. Registries may choose to record this locally if they can follow-up all babies to ascertain whether the testis descends normally.
Unspecified ectopic testis
Vesico-ureteral reflux. Registries should record and transmit to EUROCAT the underlying anomaly, if present.
Clicking hip
Clubfoot where this is no further specification of whether malformation or postural origin