EUROCAT GUIDE 1

FOR THE REGISTRATION OF CONGENITAL ANOMALIES

Edited by

Philippe DE WALS
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An E.E.C. Concerted Action Project

Department of Epidemiology,

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FOREWORD

This guide-book is intended for use in all the Centers participating in the EUROCAT Project of the E.E.C. on the registration of congenital anomalies. It represents both a synthesis and an up-dating of three previous publications : (1) EUROCAT Guide-lines for Registration of Congenital Abnormalities and Multiple Births, 1980, by J.A.C. Weatherall, (2) EUROCAT Guide-lines for Tabulation of Congenital Abnormalities and Multiple Births, 1981, by J.A.C. Weatherall and P. De Wals, (3) EUROCAT Guide-lines for Data Security, 1981, by P. De Wals and J.A.C. Weatherall. The authors are indebted to Ms. S. Goyens, Ms. D. Mousset, Dr. E. Terzian, Dr. J. Little, Ms. H. Dolk and Mr. Y. de Kettenis who helped in the preparation of this guide.

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CONTENTS

I. OBJECTIVES OF THE EUROCAT PROJECT

The EUROCAT Project is a Concerted Action of the European Economic Communities (E.E.C.) in the field of the epidemiology of congenital anomalies. The Concerted Action Programme was established by the Council decision of 13th February 1978[1]. It has been continued as part of the Medical Research Programme on Pre-, Peri-, and Postnatal Care[2].

The long term objective of the EUROCAT Project is to test the feasibility of carrying out epidemiological surveillance in the countries of the E.E.C. taking congenital anomalies as an example.

The specific objectives of the study are :

(1) to establish in each country of the E.E.C. one or several regional registers of congenital anomalies providing reliable epidemiologic information on the occurrence of the registered conditions in the progeny of the population living in a defined geographic area;

(2) to harmonize the methods of diagnosis and of data collection in order to ensure the comparability of statistics between participating centres;

(3) to monitor the occurrence rate of congenital anomalies in the progeny of different population groups in order to identify any unexpected frequency;

(4) to investigate the risk associated with possible teratogenic factors;

(5) to create in each country an area where reporting is reliable, so that base line rates are available for calibrating any information system established at national level for the detection of adverse environmental influences;

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1. OJ No L52, 23.02.1978, p 20.
2. OJ No L248, 24.08.1978, p 15.

(6) to evaluate the effectiveness and efficiency of screening programmes, of preventive measures, and of treatment methods;

(7) to provide a well documented set of cases recorded in a defined population for clinical research.

The registration of congenital anomalies actually began in 1979 with the participation of 12 centres. In 1984, 17 centres from the 10 countries of the E.E.C. are collaborating in the Project (list in Appendix I). Two centres from countries outside E.E.C. are also participating in the registration. Other centres within or outside the E.E.C. could possibly join the study.

Each EUROCAT Register is directed by a Registry Leader (list in Appendix I) who is responsible for day to day running of the regional register. The financing of the activities in each Register is the responsibility of the Member States of the E.E.C.

The coordination of the EUROCAT Project is financed by the Commission of the E.E.C. and is carried out by a Project Leader with supporting staff. The coordinating centre (EUROCAT Central Register) is situated in the Department of Epidemiology, School of Public Health, University of Louvain, Brussels. The scientific supervision of the Project is the responsibility of the COMAC-Epidemiology of the E.E.C.

II. INFORMATION TO BE RECORDED AT LOCAL LEVEL

Ideally, every EUROCAT Register should record any congenital anomaly occurring in any conception in women resident in a geographically defined area. This involves searching for structural defects (congenital malformations), chromosomal anomalies, metabolic disorders, and any heriditary disease in aborted fetuses, in live and stillborn infants, in dead children, and during childhood.

When it is not possible to achieve a complete registration of birth defects in the total births to mothers resident in a geographically defined area, the total number of births to mothers whether resident or not, taking place in that area, can be used as reference population.

As there is no general agreement about the definition of congenital anomaly, local physicians should be asked to notify to their register anything they consider to be a congenital malformation, a chromosomal anomaly, a metabolic disorder, or a hereditary disease. This is recommended with the intention of making the detection as sensitive as possible even if trivial conditions are over-reported. It is easier to exclude notified cases with trivial conditions than to search for unreported cases of severe malformations.

There is no limit to age at registration, cases to be recorded include malformed fetuses, newborn babies, and children.

It is expected that all registers will, in the first instance, achieve a complete coverage of cases discovered at birth (live and stillborn babies) and during the early neonatal period (0-7 days). It is essential that an appropriate follow up of registered cases is established in order to confirm diagnoses and to exclude falsely reported cases.

The case-finding should also extend to all spontaneous abortions irrespective of gestational age and to all induced abortions following pre-natal diagnosis at any gestational age.

In a second stage, the registration should cover all anomalies discovered in children up to school age.

The EUROCAT registers will record the identity and address of each case. At local level, the use of named records is needed (1) to link reports arriving from several sources and so avoid a multiple registration; (2) to allow the follow up of cases for which a confirmation of the diagnosis is required; (3) to study the outcome of treatment and the survival of malformed children; (4) to trace the cases in order to conduct prospective or retrospective enquiries.

The recording of information about the case and its mother and father (Appendix II) is requested by EUROCAT, along with the diagnosis of all malformations and a local number for each case. It is considered essential that the following are included in the information : date of birth of the baby, place of birth, sex, type of birth, birth weight, length of gestation, maternal age at delivery, place of residence, every congenital anomaly observed, and time of discovery.

It is understood that some centres have a special interest in particular topics and are recording other specific information in addition to the information needed for EUROCAT purposes.

III. INFORMATION TO BE TRANSMITTED TO THE CENTRAL REGISTRY

All the cases recorded at local level with single or multiple malformations included in chapter 14 of the International Classification of Diseases, 9th Edition, vol. 1, or named "congenital", "inherited", "inborn error" in other chapters, are to be transmitted to the EUROCAT Central Registry.

Since some minor and isolated malformations are considered to be unimportant, it is recommended not to transmit the cases included in the list of exclusion (Appendix IX).

The cases for which the diagnosis of malformation is well established are to be transmitted to the Central Registry regularly. The cases which are certainly malformed but for which the exact diagnosis is not yet certain should also be transmitted. When further information is available about a case already notified, an amending report is to be transmitted. The cases for which a malformation is only suspected are to be recorded locally and transmitted only when the diagnosis is confirmed by follow-up.

For the transmission of information about eligible cases, a standard pre-coded transmission form (Appendix II) and the appropriate coding instructions are shown in Appendix III. If local registers are using different codes for their data, the Central Registry must be informed.

The names and addresses of cases and hospitals are to be kept at the local register department and are not sent to the Central Register. The local identity number, unique for each baby, is to be transmitted.

For the written information in the completed form, the use of French or English is requested.

It is possible to send data using magnetic tapes or diskets provided that both the material and the recorded information are compatible with the EUROCAT requirements (Appendix III), and that the data have been validated according to the criteria in Appendix VI. For the transmission

of computerized information, the layout of the EUROCAT Central File as described in Appendix V should be used for formating local data.

IV. RECOMMENDATIONS FOR TABULATION

The purpose of this section is to describe some methods used for the tabulation of congenital anomalies at the EUROCAT Central Register. In order to achieve comparability in published statistics, it is recommended to each participating centre to adopt similar rules.

Terminology

The terminology used in EUROCAT tabulations is that of the British Paediatric Association Classification of Diseases (1979). Any condition presented in a table is defined according to its corresponding code number that could be at the level of three, four, or five digits. A sixth digit could be used should the need arise.

Classification

For routine tabulation, the congenital anomalies reported in registered infants are classified according to three levels :

(1) a list of selected defects which are clinically or etiologically defined entities is used for routine tabulation (Appendix VIII). This list is compatible with the lists used by the United States Birth Defect Monitoring Program and by the International Clearinghouse for Birth Defects Monitoring Systems. Babies having none of the selected conditions are not included in this tabulation;

(2) congenital anomalies are classified into 38 categories or subgroups (Appendix VII). This second list includes all possible conditions. Each baby registered is included at least somewhere in this tabulation;

(3) the 38 subgroups are classified into 15 categories or groups (Appendix VII). This set includes all possible conditions and each baby is counted at least once in the table. This list is compatible with the list used by the Congenital Malformation Notification System of the Office of Population Census and Survey, in London.

In all these tabulations, a baby with one or several conditions falling into different categories, is counted once within each class. The numbers in classes cannot be added to reach a total of babies.

In addition, separate tabulations should be made for babies with a single anomaly and those with more than one anomaly.

Type of birth

Four types of birth are considered : spontaneous abortion, induced abortion, stillbirth, and live birth. The definitions recommended by EUROCAT are those of the World Health Organization (Appendix IV).

In every published table the types of birth which are included in figures should be indicated. Prevalence rates of congenital anomaly at birth should be expressed by the number of affected individuals divided by the total number of births (livebirths and stillbirths). For the analysis of period prevalence rates in cohort, total births during a given period of time should be used as reference population.

Time of diagnosis

For many conditions, the period during which babies are observed is an important determinant of the reported prevalence rate. As a result, the upper limit of the observation period should be indicated in all tabulations.

For live born babies, the periods of observation concerning the time of diagnosis should be grouped as follows : (a) prenatally, at birth or within seven days of birth, (b) after seven days of birth and before one year of age, (c) after one year of age.

V. GUIDE-LINES FOR DATA SECURITY

The fulfilment of the objectives of the EUROCAT project implies, for each participating centre, the recording of the identity of malformed babies and their parents along with a list of social and medical private information. At local level the use of named records is needed (1) to link reports arriving from several sources, so avoiding multiple registrations; (2) to allow the follow up of cases which is necessary to ascertain the diagnosis and to study the outcome of malformed children; (3) to trace the cases in order to conduct prospective or retrospective etiological studies; (4) to ensure the delivery of all possible aid to the malformed children and their families.

The keeping of such a register raises at local level the problems of privacy, confidentiality and acceptability to the concerned families and to the population. It is thus essential that local conventions and/or national regulations must be respected by each local register. The local register is kept under the responsibility of the Registry Leader or the Head of the Academic Department concerned.

In order to perform epidemiological surveillance, data collected by each participating centre are centralized. The EUROCAT Central Register is presently located in the Department of Epidemiology, at the Catholic University of Louvain, in Brussels, Belgium. As this procedure involves the transmission of data over country borders, the strongest precautions are taken against any unauthorized use of the data bank and against any breach of confidentiality.

At the present time, there is no specific data protection act in Belgium and no special recommendations have been issued by the E.E.C. However, the Assembly of the European Science Foundation issued on November 12th, 1980 a statement concerning the protection of privacy and the use of personal data for research which lays down general principles. The EUROCAT Guide-Lines are considered to be in agreement with these recommendations.

The adopted procedure is as follows :

1. At local level , personal data, defined as any information relating to an identified or identifiable individual, are guarded at the local registry department under the responsibility of the Registry Leader or the Head of the Department.

2. Information about each case that is to be transmitted to the EUROCAT Central Register is stated in Appendix III.

3. No name or detailed address or detailed location of birth are transmitted to the EUROCAT Central Registry. Most of the personal data are already coded (Appendix III). Local codes are used for designating places of birth and places of residence. These codes used in transmitted records should be for sufficiently large groups so that individual cases cannot be identified at the Central Register.

4. From each local Register, data are transmitted to the Central Register by using either the EUROCAT Transmission Form (Appendix II), or magnetic tapes containing coded information.

5. Transmission forms or magnetic tapes are sent in batches by mail.

6. At the Central Registry, the transmitted local forms and magnetic tapes are retained in locked cabinets. Access is only available to EUROCAT Register Staff.

7. The transmitted data are entered into the computer whose magnetic storage units are located within the department. A copy of the computerized file is retained in a locked cabinet.

8. The EUROCAT Central File is described in Appendix V. Access to the file is restricted to the Registry Staff members. A password is used.

9. No personal and medical information concerning individual cases will be released other than to the register from which the data was

sent. For research use centrally, only statistical data will be available and only through the staff in charge of the Register.

10. For the EUROCAT file on the central computer, there is no way in which an individual can be identified and the EUROCAT Register cannot be linked with any other register.

11. If specific enquiries and researches concerning individual cases and needing additional information are to be conducted, the project will be first submitted to the Project Management Group. Such studies will usually start by identifying cases in the Central Registry. Further information will be obtained by reference to the original local form held at the Central Registry. From thence enquiries will be made which will refer to the local registry and may involve reference to medical notes or to attendant physicians. An approach to individual families will be asked for only if no other means can be used and if the study is considered to be of sufficient importance by the Project Management Group and local ethical committees if such exist.

12. Transmitted data will be retained at the Central Register for at least 5 years from the date of receipt when a decision will be made on the merits of retaining them for longer.

13. At the EUROCAT Central Registry, the security of data is under the responsibility of the Project Leader and of the Head of the Department of Epidemiology.

VI. GUIDE-LINES FOR PUBLICATION

Two types of publication related to the EUROCAT Project are considered : those arising from one or several local registers, and those made at the initiative of the Central Registry.

Local registers are encouraged to publish papers and reports on special aspects of their findings. Any local publication related to the EUROCAT Project should mention the collaborative character of the study : The EUROCAT Project is a Concerted Action of the European Economic Communities. Such a publication should be transmitted to the Project Leader for his information. Any paper with comparative content based upon the unpublished data of other participating centres cannot be published without the agreement of the respective centres.

The collaborative nature of the project implies that the data transmitted to the Central Register are not the property of any one centre or individual, but belong to all involved in the project under the aegis of the E.E.C. Any publication made at the initiative of the Central Registry should be approved by the Editorial Board of the EUROCAT Project, to which members are appointed by the COMAC-EPIDEMIOLOGY of the E.E.C. The Editorial Board has authority concerning all the problems arising in publication policy.

For autorship, the principle investigator(s) of any particular study should be the first author(s). Registry Leaders of participating centres should be coauthors in alphabetical order.

APPENDIX I : LIST OF PATICIPATING CENTRES AND REGISTRY LEADERS

NAME OF REGISTER	COUNTRY	REGISTRY LEADER	ADDRESS
WEST-FLANDERS	BELGIUM	Prof. R. BECKERS	Ministerie van Volks-gezondheid en van het Gezin Administratief Centrum Vesaliusgebouw B - 1010 BRUSSEL Tel : (2) 564.80.11
HAINAUT	BELGIUM	Dr. I. BORLEE	Ecole de Santé Publique U.C.L.- EPID 30.34 Clos Chapelle-aux-Champs 30 B - 1200 BRUXELLES Tel : (2) 764.33.20
ODENSE	DENMARK	Dr. M. ULRICH	Institute of Medical Genetics Odense University J.B. Winslows VEJ 17 DK - 5000 ODENSE C Tel : (9) 11.33.33 ext.4944
PARIS	FRANCE	Dr. J. GOUJARD	Groupe de Recherches Epidémiologiques sur la Mère et l'Enfant INSERM Av. P.V. Couturier 16 bis F - 94800 VILLEJUIF Tel : (1) 677.24.69
STRASBOURG	FRANCE	Prof. C. STOLL	Institut de Puériculture Hospice Civil de Strasbourg 23 rue de la Porte de l'Hôpital BP N° 426 F - 67000 STRASBOURG Tel : (88) 36.71.11 ext.3048
WEST-BERLIN	GERMANY	Dr. G. KARKUT	Frauenklinik und Poliklinik im Klinikum Steglitz der Freien Universität Berlin Hindenburgdamm 30 D - 1000 BERLIN 45 Tel : (49) 30/798.25.93

EVIA	GREECE	Dr. S. TSAGARAKI	Children's Hospital Agia Sophia Institute of Child Health GR - ATHENS 617 Tel : (1) 770.92.41
EMILIA ROMAGNA	ITALY	Dr. E. CALZOLARI	Istituto di Genetica Medica Via L. Borsari 46 I - 44.100 FERRARA Tel : (532) 35021
FIRENZE	ITALY	Dr. C. GALANTI	Regione Toscana Dipartimento Sicurezza Sociale 26 Via di Novoli I - FIRENZE CAP 50100 Tel: (55) 439.32.75
UMBRIA	ITALY	Dr. A. CALABRO	Clinica Pediatrica University di Perugia Monteluce I - 06100 PERUGIA Tel : (75) 25913
DUBLIN	IRELAND	Dr A. RADIC	The Medico-Social Research Board 73 Lower Baggot Street IRL - DUBLIN 2 Tel : (1) 76.60.76
GALWAY	IRELAND	Dr. D.F. LILLIS	Regional Hospital Dpt. of Paediatrics IRL - GALWAY Tel : (91) 64141/5
LUXEMBURG	LUXEMBURG	Dr.D.HANSEN-KOENIG	Ministère de la Santé Publique Division de Médecine Préventive et Sociale 22 rue Goethe L - 1637 LUXEMBOURG Tel : (352) 40801
GRONINGEN	NETHERLANDS	Dr. L.P. TEN KATE	Dpt. of Human Genetics University of Groningen 4 Ant. Deusinglaan NL - GRONINGEN Tel : (50) 11.61.20
GLASGOW	UNITED KINGDOM	Dr. F. HAMILTON	Greater Glasgow Health Board 225 Bath Street GB - GLASGOW G2 4JT Tel : (41) 332.29.77 ext.118

LIVERPOOL	UNITED KINGDOM	Dr. F. HARRIS	Institute of Child Health Alder Hey Children's Hospital Eaton Road GB - LIVERPOOL L12 2AP Tel : (051) 228.4811
BELFAST	UNITED KINGDOM	Prof. N. NEVIN	The Queen's University of Belfast Dpt. of Medical Genetics Institute of Clinical Science Grosvenor Road GB - BELFAST BT 12 6BJ Tel : (232) 24.05.03 ext.2638/2688

Associate centers not in E.E.C. countries

LAUSANNE ZURICH	SWITZERLAND	Prof. T. PEXIEDER	Inst. d'Histologie et d'Embryologie 9 rue du Bugnon CH - 1011 LAUSANNE Tel : (21) 23.22.92
ZAGREB	YUGOSLAVIA	Prof. I. SVEL	Zavod za Zastitu Majki i Djece 41000 ZAGREB, KLAICEVA 16 YUGOSLOVIA

APPENDIX II

TRANSMISSION FORM

APPENDIX III : INSTRUCTIONS FOR CODING

General instructions : One form to be completed for each abnormal baby or fetus.
In general, the code "8" should be used for "other", and
the code "9" should be used for "not known".
An asterisk (*) indicates the items considered to be essential for the Central Registry.

ITEM	DIGITS	CODE	INSTRUCTIONS	BOX NO.
DATA ABOUT THE BABY				
Local Identification number *	11	Local code		-
EUROCAT serial number	11	Registry code number 01 : W. Vlaanderen (B) 02 : Hainaut (B) 03 : Odense (DK) 05 : Paris (F) 07 : W. Berlin (D) 08 : Firenze (I) 09 : Umbria (I) 10 : Dublin (IRL) 11 : Galway (IRL) 12 : Luxembourg (L) 13 : Groningen (NL) 14 : G. Glasgow (UK) 15 : Liverpool (UK) 16 : Belfast (UK) 17 : Eubia (GR) 18 : Emilia Romagna (I) 19 : Strasbourg (F) 20 : Zurich (CH) 21 : Zagreb (YU)	Enter the code allocated to you in the first two boxes. The remaining boxes will be completed by Central Registry.	1-2 3-11
Block reference	1	1	Already preprinted on form	12
Date of birth *	6		Day/Month/Year e.g. 21st July 1980 code 2 1 0 7 8 0	13-18
Place of birth *	5	99999 = Not known	Use local code for identifying institutions or other places of birth such as home delivery.	19-23

ITEM	DIGITS	CODE	INSTRUCTIONS	BOX NO.
Sex *	1	1 : Male 2 : Female 3 : Indeterminate 9 : Not known		24
Number of babies/ fetuses delivered	1	1 : Singleton 2 : Twins 3 : Triplets...etc. . . 9 : Not known	Complete one form for each malformed baby. Only one form to be completed for conjoined twins.	25
Birth order (in multiple set)	-	-	To be completed in writing for multiple delivery only : 1, 2, 3,...	-
Type of birth *	1	1 : Live birth 2 : Stillbirth 3 : Spontaneous abortion 4 : Induced abortion 9 : Not known	See definitions in Appendix IV.	26
Birth weight *	4	9999 : Not known	Complete the weight in grams. See Appendix IV.	27-30
Date of last menstrual period	6	999999 : Not known 000000 : No L.M.P.	Day/Month/Year e.g. 21st July 1980 2 1 0 7 8 0	31-36
Length of gestation *	2	99 : Not known	In completed weeks. If L.M.P. is available and certain, calculate gestational age from data of L.M.P. to date of delivery (Appendix IV).	37-38

ITEM	DIGITS	CODE	INSTRUCTIONS	BOX NO.
			If L.M.P. is available but uncertain, give the corrected gestational age by clinical ascertainment or other means. If L.M.P. is not available, give the estimated gestational age by clinical ascertainment or other means.	
Certainty of L.M.P.	1	1 : certain 2 : uncertain 3 or 0 : no L.M.P. 9 : not known		39
Amniocentesis	1	1 : performed, result known 2 : performed, result unknown 3 or 0 : not performed 8 : failed 9 : not known	If performed, record indication(s), investigation(s), and results in the space for comments, etc. or on the back.	40
Ultrasound	1	1 : performed, result known 2 : performed, result unknown 3 or 0 : not performed 9 : not known	As for box 40	41
Other techniques	1	1 : performed, result known 2 : performed, result unknown 3 or 0 : not performed 9 : not known	If performed, specify technique and give results in the space for comments.	42
Karyotype of infant/ fetus	1	1 : performed, result known 2 : performed, result unknown 3 or 0 : not performed 9 : not known	If performed, record the results in the space provided.	43

ITEM	DIGITS	CODE	INSTRUCTIONS	BOX NO.
Post mortem examination	1	1 : performed, result known 2 : performed, result unknown 3 or 0 : not performed 9 : not known	If performed, record the malformation(s) discovered in the "Malformations" section in the form. If other findings record on the back of the form or in the "comments" space.	44
Source of information	5 (max.)	1 : Notes in routine scan 2 : Birth notificaiton or notification of malformation at birth 3 : Hospital case notes 4 : Death or stillbirth certificate 5 : Pre-natal diagnosis report 6 : Laboratory report (cytogenetic, chemical, etc.) 7 : Post mortem examination 8 : Other 9 : Not known	Indicate each source of information - up to a maximum of five - used in collecting information on the child.	45-49
Date of death	6	000000 : Alive 999999 : Not known whether alive or dead	Day/Month/Year (as for 13-18) Write clearly whether the child is known to be dead, known to be alive or whether it is not known if the child is dead or alive, at the age of 7 days, one month, and one year. Give day, month, and year as best known (e.g. 99 05 82 means dead in May 82).	50-55
Previous sibs notified to Central Registry	1	1 : yes 2 : no 9 : not known		56

ITEM	DIGITS	CODE	INSTRUCTIONS	BOX NO.
Local code number	-		Enter the local identification number notified to Central Registry - for family linkage purposes.	
Number of sibs with same type of anomaly	1	0 : none 9 : not known	Enter number, e.g. "1" for one sibling affected by same type of anomaly".	57
Number of sibs with other anomalies	1	0 : none 9 : not known	Enter number, e.g. "1" for one sibling affected by other type of anomaly	58
Mother - same type of anomaly	1	1 : yes 2 : no 9 : not known	If Mother is similarly affected enter "1" for "yes" and give description in "comments" space or on back of form.	59
Father - same type of anomaly	1	1 : yes 2 : no 9 : not known	As for box 59	60
DATA ABOUT THE MOTHER				
Block Reference	1	2	Already preprinted on form.	12
Residence code *	7	9999999 : unknown	Use local code for locality (area) of residence.	13-19
Date of birth	6	999999 : if not known or not available.	Day/Month/Year e.g. 1st October 1945. Give as much of data as is available. code 0 1 1 0 4 5	20-25
Age at delivery *	2	99 : if not known	In completed year at the time of delivery.	26-27
Number of spontaneous abortions	1	9 : if not known	If a twin pregnancy aborted, code as 2. See definitions in Appendix IV.	28

ITEM	DIGITS	CODE	INSTRUCTIONS	BOX NO.
Number of induced abortions	1	9 : if not known	As for box 28	29
Number of live births	2	99 : if not known	A twin delivery counts as 2 if both live born, triplets as 3, etc.	30-31
Number of stillbirths	1	9 : if not known	A twin delivery counts as 2 if both stillborn, triplets as 3, etc.	32
Total number of previous pregnancies	2	99 : if not known	This is total previous pregnancies, not births. The present notified pregnancy is not to be included. Include miscarriages (abortions). Twin or other multiple pregnancies count as 1 in the total, e.g. : 3 previous LB + 1 termination of a twin pregnancy = 0 4	33-34
Occupation	5	99999 : unknown	If possible, code according to the International Classification of Occupations (ILO code). Give as much detail as possible. Give main occupation at the time of conception. Give job, industry, status in verbal description, if possible (English or French).	35-39
Social status	-	9 : unknown	Use local code	40
Racial type	-	9 : unknown	Use local code	41
Chronic illness	1	1 : yes 2 or 0 : no 9 : not known	Indicate illness(es) and give week(s) of gestation	42

ITEM	DIGITS	CODE	INSTRUCTIONS	BOX NO.
Illness during pregnancy (acute)	1	1 : yes 2 or 0 : no 9 : not known	Indicate illness(es) and give week(s) of gestation	43
Smoking	1	1 : yes 2 or 0 : no 9 : not known	"Yes" means is smoker and history is available. "No" means is not smoker and history is available. "Not known" means no history is available.	44
Alcohol	1	1 : yes 2 or 0 : no 9 : not known	As in box 44	45
Drugs during pregnancy	1	1 : yes 2 or 0 : no 9 : not known	Indicate which types and when administered	46
Consanguinity	1	1 : 1st cousin 2 : 2nd cousin 8 : other relation or not stated 9 : not known 0 : not related		47
DATA ABOUT THE FATHER			See same items under "Mother" and use the same codes.	48-66
DATA ABOUT THE MALFORMATIONS				
Block reference	1	3	Already preprinted on form.	12

ITEM	DIGITS	CODE	INSTRUCTIONS	BOX NO.
Date of discovery	6	999999 : not stated	The date on which the baby was first suspected or recognized as being malformed, even if the detailed diagnosis is not available. Day/Month/Year e.g. 14th January 1980 code 1 4 0 1 8 0	13-18
When discovered *	1	1 : birth 2 : less than 1 week 3 : 1-4 weeks 4 : 1-12 months 5 : over 12 months 6 : pre-natal diagnosis 7 : spontaneous or induced abortion 8 : at post mortem 9 : not known		19
Condition at discovery	1	1 : live 2 : dead 9 : not known		20
Syndrome, eponym or disease name *	6	ICD 9th revision code or modifications stated in next column.	Use ICD 9th revision code in the first four digits, the British Paediatric Association Classification of Diseases in the fifth digit, and the EUROCAT Guide to Eponyms and Syndromes in the sixth digit. If not a recognizable syndrome, eponym, or disease name leave blank.	21-26

ITEM	DIGITS	CODE	INSTRUCTIONS	BOX NO.
Syndrome, etc. contd.			If a recognizable syndrome, eponym, or disease name with no specific code, then code 888888. It is recommended that all the anomalies observed by the local clinician should be coded in addition to the specific name. For the written descriptions, please use English or French. If only the name of the specific syndrome, eponym, or disease is available, code in boxes 21-26 and leave the remainder blank (i.e. boxes 27-74). e.g. Trisomy 18/Edwards syndrome, code in boxes 21-26 7 5 8 2 0 0 When two syndromes are present in the same subject, code the more important in 21-26 and the other in 27-32.	
Malformations present *	6	BPA code	When a specific syndrome, eponym, or disease is reported in 21-26, list the observed anomalies from box 27 on. These anomalies should be coded as if they are isolated anormalies in the BPA code.	27-74

ITEM	DIGITS	CODE	INSTRUCTIONS	BOX NO.
Malformations present contd.			e.g. congenital rubella embryopathy with bilateral cataracts and unspecified abormality of heart code as 7 7 1 0 - 0 in 21-26 and in boxes 27-32 bilateral cataracts 7 4 3 3 2 and in 33-38 unspecified heart abnormality 7 4 6 9 9 Do not indicate multiple minor anomalies which are part of a well established named specific syndrome, eponym or disease. e.g. Down's syndrome with a ventricular septal defect and Brushfield spots. Enter Down's syndrome 7 5 8 0 9 0 in 21-26 and enter ventricular septal defect 7 4 5 1 4 in 27-32, but omit Brushfield spots.	
Confirmation of diagnosis	1	1 : certain 2 : still doubtful 3 : lost to follow-up 9 : not known 0 : no malformation	In cases for which the diagnosis of malformation is well established - transmit monthly, together with those for which the exact diagnosis is not yet confirmed. When this is confirmed or further information is available an amended report should be sent. In cases where a malformation is only suspected these should be recorded locally and sent to EUROCAT only when the diagnosis is confirmed by appropriate follow-up.	75

ITEM	DIGITS	CODE	INSTRUCTIONS	BOX NO.
Mode of inheritance	1	0 : autosomal dominant 1 : autosomal recessive 2 : X-linked recessive/ dominant 3 : numeric chromosomal defect of autosomes 4 : structural chromosomal defect of autosomes 5 : numeric sex chromosomal defects 6 : structural sex chromosomal defects 7 : polygenic/multifactorial 8 : known environmental agent 9 : other or unknown	optional	76
(For local use)	1		For local use	77

APPENDIX IV : RECOMMENDED DEFINITIONS

1. Live birth

Live birth is the complete expulsion or extraction from its mother of a product of conception, irrespective of the duration of the pregnancy, which, after such separation, breathes or shows any other evidence of life, such as beating of the heart, pulsation of the umbilical cord, or definite movement of voluntary muscles, whether or not the umbilical cord has been cut or the placenta is attached; each product of such a birth is considered live born.

2. Stillbirth

Stillbirth is the complete expulsion or extraction from its mother of a product of conception of 28 weeks (196 days) gestation or more, which shows no evidence of life such as breathing, spontaneously or after stimulation, beating of the heart, or pulsation of the umbilical cord, or when post mortem examination fails to show air expansion in the lungs.

3. Abortion

Abortion is the expulsion, complete or incomplete, or extraction of the product of conception prior to 28 completed weeks of pregnancy (up to 195 days) which shows no evidence of life such as breathing spontaneously, beating of the heart, pulsation of the umbilical cord, or definite movement of voluntary muscles.

4. Birthweight

The first weight of the fetus or newborn obtained after birth. This weight should be measured preferably within the first hour of life before significant postnatal weight loss has occurred.

5. Gestational age

The duration of gestation is measured from the first day of the last normal menstrual period. Gestational age is expressed in completed days or completed weeks (e.g. events occurring 280 to 286 days after the onset of the last normal menstrual period are considered to have occurred at 40 weeks of gestation).

APPENDIX V

LAYOUT OF THE CENTRAL FILE

Remark : For the transmission of computerized material to the EUROCAT Central Registry, it is recommended to use the format described in this appendix.

ITEM	VAR NO.	VAR LABEL	FORMAT	COL.	VALUE FOR BLANK	MISSING VALUE VALUES	LABEL
-	001		N 6	1-6		for Central use	
-	002		N 2	7-8	-	for Central use	
Local No	003	LOCAL No	AN 11	9-19	0	00000000000 99999999999	
Registry code number	004	REGISTER	N 2	20-21	0	00 99	01=W.VLAAND 02=HAINAUT 03=ODENSE 05=YVELINES 07=W.BERLIN 08=FIRENZE 09=ROMA 10=DUBLIN 11=GALWAY 12=LUXEMB. 13=GRONINGEN 14=GLASGOW 15=LIVERPOOL 16=BELFAST 17=EVIA 18=EM.ROMA. 19=STRASBOURG 20=ZU-LAUSANNE 21=ZAGREB
Year	005	YEAR	N 2	22-23	0	00 99	
Eurocat Serial No	006	EUROCAT No	N 4	24-27	0	0000 9999	
Date of birth day	007	BIRTH-DAY CASE	N 2	28-29	0	99 00	
Date of birth month	008	BIRTH-MONTH CASE	N 2	30-31	0	99 00	
Date of birth year	009	BIRTH-YEAR	N 2	32-33	0	99 00	

ITEM	VAR NO.	VAR LABEL	FORMAT	COL.	VALUE FOR BLANK	MISSING VALUES	VALUE LABEL
Place of birth	010	BIRTH PLACE	N 5	34-38	0	00000 99999	
Sex	011	SEX	N 1	39	9	9	1 = MALE 2 = FEMALE 3 = INDETER. 9 = NK
Number of babies	012	BABIES NO	N 1	40	9	9	
Birth order	013	BIRTH ORD	N 1	41	0	0 9	
Type of birth	014	BIRTH TYPE	N 1	42	9	9 0	1 = LB 2 = SB 3 = SA 4 = IA 9 = NK = 0
Birth weight (Field 15+16)	015	WEIGHT	N 4	43-46	9	9999 0000	
Date of LMP day	016	LMP DAY	N 2	47-48	0	99 00	
Date of LMP month	017	LMP MONTH	N 2	49-50	0 00	99	
Date of LMP year	018	LMP YEAR	N 2	51-52	0	99 00	
Length of gestation	019	GESTATION LENGTH	N 2	53-54	9	00 99	
Certainty of LMP	020	CERTAINTY LMP	N 1	55	9	9	1 = CERTAIN 2 = UNCERTAIN 0 = NO L.M.P. 3 = NO L.M.P. 9 = NK
Amnio-centesis	021	AMNIOCENT	N 1	56	9	9	0 = 3 = NO 1 = YES RK 2 = YES RNK 9 = NK

ITEM	VAR NO.	VAR LABEL	FORMAT	COL.	VALUE FOR BLANK	MISSING VALUES	VALUE LABEL
Ultra-sound	022	ULTRASO	N 1	57	9	9	0 = 3 = NO 1 = YES RK 2 = YES RNK 9 = NK
Other tech-niques	023	PREDIAG	N 1	58	9	9	0 = 3 = NO 1 = YES 2 = 9 = NK
Karyotype	024	KARYOTYPE	N 1	59	9	9	0 = 3 = NO 1 = YES RK 2 = YES RNK 9 = NK
Post-mortem exami-nation	025	POST-MORTEM	N 1	60	9	9	0 = 3 = NO 1 = YES RK 2 = YES RNK 9 = NK
First source of infor-mation	026	INFOR 1	N 1	61	9	9	1 = ROUTINE 2 = BIR-CERT 3 = HOSPITAL 4 = DEATHCER 5 = PRENATAL 6 = LAB-REP 7 = PM REP 8 = OTHER 9 = NK 0 = NO
Second source of infor-mation	027	INFOR 2	N 1	62	9	9	as above
Third source of infor-mation	028	INFOR 3	N 1	63	9	9	as above
Fourth source of infor-mation	029	INFOR 4	N 1	64	9	9	as above
Fifth source of infor-mation	030	INFOR 5	N 1	65	9	9	as above

ITEM	VAR NO.	VAR LABEL	FORMAT	COL.	VALUE FOR BLANK	MISSING VALUES	VALUE LABEL
Date of death day	031	DATE DEATH DAY	N 2	66-67	0	00 99	
Date of death month	032	DATE DEATH MONTH	N 2	68-69	0	00 99	
Date of death year	033	DATE DEATH YEAR	N 2	70-71	0	00 99	
Age at death	034	AGE DEATH	N 4	72-75	0	9999	
Status at 7 days	035	STATUS 7	N 1	76	9	9	1 = ALIVE 2 = DEAD 9 = NK
Status at 28 days	036	STATUS 28	N 1	77	9	9	1 = ALIVE 2 = DEAD 9 = NK
Status at 365 days	037	STATUS 365	N 1	78	9	9	1 = ALIVE 2 = DEAD 9 = NK
Previous sibs notified	038	PREVIOUS SIBS	N 1	79	9	9	
No of sibs with same anomaly	039	SAME ANOMALY NO	N 1	80	9	9	
No of sibs with other anomaly	040	OTHER ANOMALY NO	N 1	81	9	9	
Mother with same anomaly	041	MOTHER ANOMALY	N 1	82	9	9	
Father with same anomaly	042	FATHER ANOMALY	N 1	83	9	9	

ITEM	VAR NO.	VAR LABEL	FORMAT	COL.	VALUE FOR BLANK	MISSING VALUES	VALUE LABEL
Residence code	043	RESIDENCE MOTHER	AN 7	84-90	0	9999999 0000000	
Date of birth day	044	BIRTH-DAY MOTHER	N 2	91-92	0	99 00	
Date of birth month	045	BIRTH-MONTH MOTHER	N 2	93-94	0	99 00	
Date of birth year	046	BIRTH-YEAR MOTHER	N 2	95-96	0	99 00	
Age at delivery	047	AGE MOTHER	N 2	97-98	9	99 00	
Previous pregnancies SA	048	SA	N 1	99	9	9	
Previous pregnancies IA	049	IA	N 1	100	9	9	
Previous pregnancies LB	050	LB	N 2	101-102	0	99	
Previous pregnancies SB	051	SB	N 1	103	9	9	
Total pregnancies	052	TOTAL PREGNANCIES	N 2	104-105	0	99	
Occupation	053	OCCUPATION MOTHER	N 5	106-110	0	99999 00000	
Social status	054	SOCIAL STATUS M	N 1	111	9	9	
Racial type	055	RACE M	N 1	112	9	9	
Chronic illness	056	CHRONIC ILLNESS M	N 1	113	9	9	1 = YES 0 = 2 = NO 9 = NK

ITEM	VAR NO.	VAR LABEL	FORMAT	COL.	VALUE FOR BLANK	MISSING VALUES	VALUE LABEL
Illness during pregnancy	057	ILLNESS PREGNANCY	N 1	114	9	9	1 = YES 0 = 2 = NO 9 = NK
Smoking	058	SMOKING M	N 1	115	9	9	as above
Alcohol	059	ALCOHOL M	N 1	116	9	9	as above
Drugs	060	DRUGS M	N 1	117	9	9	as above
Consan-guinity	061	CONSAN-GUINITY	N 1	118	9	9	1 = FIRST C. 2 = SECOND C. 8 = OTHER 0 = NOT REL. 9 = NK
Date of birth day	062	BIRTH DAY FATHER	N 2	119-120	0	99 00	
Date of birth month	063	BIRTH MONTH FATHER	N 2	121-122	0	99 00	
Date of birth year	064	BIRTH YEAR FATHER	N 2	123-124	0	99 00	
Age at birth	065	AGE FATHER	N 2	125-126	9	99 00	
Occu-pation	066	OCCUPATION F.	N 5	127-131	0	99999 00000	
Social status	067	SOCIAL STATUS F	N 1	132	9	9	
Racial type	068	RACE F	N 1	133	9	9	
Chronic illness	069	CHRONIC ILLNESS F	N 1	134	9	9	1 = YES 0 = 2 = NO 9 = NK
Smoking	070	SMOKING F	N 1	135	9	9	as above
Alcohol	071	ALCOHOL F	N 1	136	9	9	as above
Drugs	072	DRUGS F	N 1	137	9	9	as above

ITEM	VAR NO.	VAR LABEL	FORMAT	COL.	VALUE FOR BLANK	MISSING VALUES	VALUE LABEL
Date of discovery day	073	DISCOVERY DAY	N 2	138-139	0	99 00	
Date of discovery month	074	DISCOVERY MONTH	N 2	140-141	0	99 00	
Date of discovery year	075	DISCOVERY YEAR	N 2	142-143	0	99 00	
When disco-vered	076	WHEN DISCOVERED	N 1	144	9	9 0	1 = BIRTH 2 = WEEK 3 = MONTH 4 = YEAR 5 = OVER YEAR 6 = PRENATAL 7 = ABORTION 8 = AT POST MORTEM 9 = NK
Condition at disco-very	077	CONDITION AT DISCO.	N 1	145	9	9	1 = ALIVE 2 = DEAD 9 = NK
Syndrome	078	SYNDROME	N 6	146-151	0	999999 000000	
First malfor-mation	079	MALFO 1	N 6	152-157	0	999999 000000	
2nd malfo.	080	MALFO 2	N 6	158-163	0	999999 000000	
3rd malfo.	081	MALFO 3	N 6	164-169	0	999999 000000	
4th malfo.	082	MALFO 4	N 6	170-175	0	999999 000000	
5th malfo.	083	MALFO 5	N 6	176-181	0	999999 000000	
6th malfo.	084	MALFO 6	N 6	182-187	0	999999 000000	
7th malfo.	085	MALFO 7	N 6	188-193	0	999999 000000	

ITEM	VAR NO.	VAR LABEL	FORMAT	COL.	VALUE FOR BLANK	MISSING VALUES	VALUE LABEL
8th malfo.	086	MALFO 8	N 6	194-199	0	999999 000000	
Confirmation of diagnosis	087	CONFIRMATION DIAG.	N 1	200	9	9	1 = CONFIRM. 2 = DOUBT 3 = LOST 0 = NOT MALFO
Mode of inheritance	088	INHERITANCE	N 1	201	9	9	0 = AD 1 = AR 2 = XL 3 = AND 4 = ASD 5 = SND 6 = SSD 7 = POLY 8 = ENVIR 9 = NK
~~Sub-group 089~~ ~~1~~		~~SG 1~~	~~N 1~~	~~202~~	~~0~~	~~0~~	

APPENDIX VI : SCHEME FOR THE LOGICAL VALIDATION OF DATA

Remark : *The logical checks described below should be used locally when already computerized data are transmitted to the Central Registry.

*In addition to these logical checks, data should be tested for the range of acceptable values as described in the coding instructions (Appendix III).

*This list of checks should not prevent outlying available values but should be used to make sure that the outlying values are correct.

1. Sequence number within a registry cannot be duplicated.

2. If, within a registry two or more records are coded with the same values of the following variables :

   date of birth or abortion of baby or fetus
   sex
   birthweight $\pm$ 100g

   a warning will be issued that a case record may have been duplicated.

3. The year of notification cannot be earlier than the year of birth or abortion.

4. Checks on internal compatibility of date of

   birth or abortion
   LMP
   death
   birth of mother
   birth of father
   discovery of malformation

   (a) if the month is known (i.e. coded as other than 99) then the year must also be known;

   (b) if the day is known (i.e. coded as other than 99) then the month and the year must also be known.

   (c) day of birth cannot be greater than 31.
   (d) day of birth in April, June, September, or November cannot be greater than 30.

   (e) day of birth in February cannot be greater than 29.

5. Checks on intervals between dates :

   (a) If the date of the LMP is coded as known, then the date of LMP must be earlier than the date of birth or abortion.

   (b) If the date of death is coded as known, then the date of death must be the same as, or later than, the date of birth or abortion.

   (c) If the date of the mother's birth is known, then a warning will be issued if this is less than twelve years before the date of delivery or abortion.

   (d) If the date of the father's birth is known, then a warning will be issued if this is less than twelve years before the date of delivery or abortion.

   (e) If the date of discovery of anomalies in the baby or fetus is coded as known, this date must be later than the date of the LMP.

   (f) If the date of discovery of anomalies in the baby or fetus is coded as known, and either prenatal diagnosis was not performed or such a diagnostic procedure was performed but the result was unknown at the time of notification, then the date of discovery must be the same as, or later than, the date of birth or abortion.

   (g) The value of the variable " when discovered" must be compatible (1) with the calculated interval between date of birth or abortion and date of discovery and (2) with the type of birth, i.e.

      (i) if birth, cannot be discovered at abortion;
      (ii) if abortion, cannot be detected at birth or subsequently;
      (iii) cannot be detected at prenatal diagnosis if procedure not performed.

6. When there is no LMP, the date of the LMP must be left unfilled.

7. Checks on consistency between gestation and type of birth or abortion :

   (a) If type of birth is coded as still, then coded gestation must be 28 weeks or more or unknown.

   (b) If abortion is coded, then coded gestation must be less than 28 weeks or unknown.

8. Doubtful combinations of estimated gestation, birthweight and outcome as a live or still birth :

OUTCOME	ESTIMATED GESTATION	BIRTHWEIGHT
Live	< 27 weeks	> 1000 g
Live	28-31 weeks	> 2000 g
Live	32-35 weeks	≤ 3500 g
Live	36-37 weeks	≥ 500 g or ≤ 3500 g
Live	38-43 weeks	≥ 1000 g or ≤ 6000 g
Live	> 44 weeks	all birthweights
Still	< 27 weeks	
	> 44 weeks	all birthweights

On the assumption that birthweight will be more accurately recorded than gestation, it is suggested that in the above instances gestation be regarded as extremely uncertain.

Birthweight, as well as gestation, may be regarded as uncertain if a weight of 5500g or more is recorded for coded gestation of 37 weeks or less and a weight of 6000g or more for 38 weeks or more.

---

9. Sources of information and performance of pre-natal diagnosis and post mortem :

   (a) A case cannot be as ascertained from records of antenatal diagnoses unless such a procedure is recorded as performed.

   (b) Similarly, a case cannot be ascertained from records of post mortems unless such an investigation was carried out.

---

10. Codes for sources of information should not be duplicated.

---

11. Maternal age should be more than 11 years and less than 46 years.

---

12. Maternal age and previous reproductive history :

   (a) Women aged 15 or less are likely to be nulliparous and therefore a warning will be issued if a parity of one or more is coded. Because of the possibility of multiple birth, it is difficult to specify "rules of thumb" for other combinations of maternal age and parity.

   (b) For this reason, the relationship between maternal age and the total number of previous pregnancies is examined. The following combinations are considered to be implausible :

AGE	NUMBER OF PREVIOUS PREGNANCIES
15 or less	3 or more
16 - 19	7 or more
20 - 24	12 or more

---

13. Previous reproductive history :

   (a) If the total number of previous pregnancies is coded as zero, the sum of the coded numbers of births and abortions must also be equal to zero.

   (b) The total number of previous pregnancies (if coded as known) cannot be greater than the sum of the coded numbers of births and abortions.

   (c) If the total number of previous pregnancies is less than the sum of the coded numbers of births and abortions (if coded as known), verify that the mother has experienced a multiple birth or multiple abortions previously.

---

14. A baby cannot be recorded as dead at the time the malformation was first detected and also be recorded as alive at the time of notification.

---

15. If the type of event was a stillbirth or abortion, or a live birth dead by the time of notification, the confirmation of diagnosis should not be recorded as "lost to follow up".

---

16. Malformations :

   (a) Issue a warning if a malformation code other than that for a syndrome is duplicated as it is unusual for separate anomalies in the same individual to be of a type classifiable into the same code.

   (b) A blank code, i.e. a set of six blank characters, should not be inserted between two malformation codes as such a practice raises the possibility that a code has been missed.

   (c) If a malformation code is entered which is not in the directory, issue a warning. Subsequent action will be either to edit the code entered or to update the directory.

   (d) A case should not be entered if affected only by one of certain minor malformations (see Appendix IX).

   (e) A case should not be entered if : only one condition reported of either cardiac murmur or congenital heart disease without mention of cyanosis, clicking hip or if there is an unconfirmed report of congenital dislocation of the hip, varus condition of the foot, e.g. talipes equinovarus, or valgus condition of the foot, e.g. talipes calcaneovalgus.

   (f) If the 4th digit of an ICD code is coded as "9", indicating that the type of the malformation is unspecified, then the certainty of diagnosis should not be coded as confirmed.

   (g) If the ICD code for a chromosomal anomaly indicates that the karyotype is known, then the performance of karyotypic investigation should be recorded as "yes, result known". If the chromosomal investigation has been unsuccesful because of failure to culture the chromosomes, the result of the investigation would be recorded as "yes, result known".

   (h) If a malformation is of a type that usually affects one sex only (e.g. X-linked conditions, undescended testis etc), then issue a warning if the coded sex is inappropriate. It is suggested that a warning rather than an error message is issued because females have been known to be affected by, as distinct from being carriers of, X-linked disorders, e.g. cases of monozygotic female twins with Duchenne muscular dystrophy have been reported.

(i) If fetal alcohol syndrome (coded 759890, 759910 or 760760) is recorded but the mother is not recorded as having consumed alcohol during pregnancy, a warning should be issued.

(j) If twin-to-twin transfusion syndrome is recorded, the birth or abortion should be recorded as multiple.

(k) If a syndrome attributable to maternal infection is recorded, then a warning should be issued if the mother was not recorded as having been affected by an acute illness or by a chronic illness during pregnancy.

(l) If spina bifida of any type is coded, then the codes for isolated hydrocephalus (742300 - 742390) should not be entered.

The checks (m)-(q) have been specified in order to verify that the minimal diagnostic criteria of syndromes are satisfied. It was decided to computerise this process for the correlations on which checks (m)-(o) are made because substantial numbers of cases were recorded during the period 1979-1982.

(m) If Pierre Robin syndrome is recorded, then micrognathia, cleft palate and anomalies of the tongue should also be verified and recorded.

(n) If trisomy 13 without karyotypic confirmation, and only one of the triad of microphthalmia, cleft lip and palate, and polydactyly, is recorded, then a warning should be issued so that the constellation of the features can be confirmed as Patau's syndrome.

(o) If Klippel - Trenaunay - Weber syndrome is recorded, then assymetric limb hypertrophy and haemangiomata should also be verified and recorded.

(p) If congenital rubella syndrome is recorded but the triad of deafness, cataracts and cardiovascular anomaly is not recorded, the basis of the diagnosis should be verified before reporting the case.

(q) If certain rare syndromes are recorded, the malformation codes should be checked by hand in order to verify that the minimal diagnostic criteria are satisfied before reporting the case.

---

17. As only data on births and abortions with malformations have been and are computerised centrally, the confirmation of diagnosis should not be coded "no malformation".

---

18. Mother and/or father with same type of anomaly as index birth or abortion.

If the mother or father is coded as having the same type of anomaly as the index birth or abortion, this should be verified as clinically plausible by reviewing the coded diagnosis.

---

19. Previous sibs with anomalies of the same or another type.

(a) If a previous sib has been notified to EUROCAT Registry
(1) the local code numbers should be verified by hand;
(2) the sum of the numbers of sibs with the same type of anomaly and those with other anomalies must not be zero.

(b) If "previous sibs notified to EUROCAT Registry" is coded other than "no", the number of previous pregnancies must not be zero.

(c) The sum of the numbers of sibs with the same type of anomaly and those with other anomalies must not exceed the sum of the numbers of previous births (live and still) and abortions (spontaneous and induced).

(d) If it is not known whether a previous sib has been notified to EUROCAT, then issue a message instructing that the following procedure is to be carried out centrally.

The data for the appropriate centre should be scanned for records which match with that of the index event on the following essential items :

1. mother's date of birth or
   year of birth or abortion minus mother's age at delivery ± 1 year

2. father's date of birth or
   year of birth or abortion minus father's age at delivery ± 1 year

3. number of previous pregnancies less than the number coded for the index birth or abortion (provided that both are known).

The output should be ranked in descending order of the number of the following items matched :

1. maternal residence
2. maternal occupation
3. maternal chronic illness
4. maternal smoking
5. maternal alcohol consumption
6. consanguinity
7. paternal occupation
8. paternal chronic illness
9. paternal smoking
10. paternal alcohol consumption

---

20. If the confirmation of diagnosis is coded as "doubtful", and the validation checks do not reveal any error, then a copy of the coded form should be generated with the comment "Has the diagnosis been

confirmed since ?"

---

21. If any of the following variables are coded as "unknown";

    date of birth
    sex
    type of birth or abortion
    number of babies delivered
    birthweight
    maternal age at delivery
    syndrome
    malformations present

    then a copy of the coded form should be generated indicating which variable(s) have been coded as unknown together with the message "Is it possible to obtain further information on the variable(s) whose value(s) has(ve) been coded as "unknown" ?

---

22. If the source of information is a death or still birth certificate only then a copy of the coded form should be generated together with the message "This case has been coded as ascertained only from a death or still birth certificate. Are information about this case available from any other source, e.g. post mortem records ?"

---

23. If an unspecified anomaly of any system is coded then a copy of the coded form should be generated together with the message "An unspecified anomaly of a particular system has been coded. Is it possible to obtain the information required to clarify the diagnosis ?"

APPENDIX VII

List of groups and subgroups of congenital anomalies classified according to the ICD 9th Revision Code and to the BPA Code.

GROUPS		SUBGROUPS		CONGENITAL ANOMALIES	
CODE		CODE		ICD CODE	
A	Nervous system	01	Dysraphic anomalies of central nervous system	7400	Anencephalus
				7401	Craniorachischisis
				7402	Iniencephaly
				7410	Spina bifida with hydrocephalus
				7419	Spina bifida without mention of hydrocephalus
				7420	Encephalocele
		02	Congenital hydrocephalus	7423	Congenital hydrocephalus without mention of spina bifida
		03	Microcephalus	7421	Microcephalus
		04	Other congenital anomalies of nervous system	7422	Reduction deformities of brain
				7424	Other specified anomalies of brain
				7425	Other specified anomalies of spinal cord
				7428	Other specified anomalies of nervous system
				7429	Unspecified anomalies of brain, spinal cord and nervous system
				3440	Quadriplegia
				3441	Paraplegia
				3442	Diplegia of upper limbs
				3443	Monoplegia of lower limb
				3444	Monoplegia of upper limb
				3445	Unspecified monoplegia
				3446	Cauda equina syndrome
				3448	Other paralytic syndrome
				3449	Other unspecified paralytic syndrome
				3492	Disorders of meninges, not elsewhere classified

GROUPS		SUBGROUPS		CONGENITAL ANOMALIES	
CODE		CODE		ICD CODE	
A	(cont.)	04	(cont.)	3519	Unspecified facial nerve disorcers
				3526	Moebius syndrome and multiple cranial nerve palsies
				225-	Benign neoplasm of brain and other parts of nervous system
				2375	Neoplasm of uncertain behaviour of brain and spinal cord
				2376	Neoplasm of uncertain behaviour of meninges
				2377	Neurofibromatosis
				2379	Neoplasm of uncertain behaviour of other and unspecified part of nervous system
				2370	Neoplasm of uncertain behaviour of pituitary gland and craniopharyngal duct
				2371	Neoplasm of uncertain behaviour of pineal gland
B	Eye	05	Congenital anomalies of eye	7430	Anophthalmos
				7431	Microphthalmos
				7432	Buphthalmos
				7433	Congenital cataract and lens anomalies
				7434	Coloboma and other anomalies of anterior segments
				7435	Congenital anomalies of posterior segment
				7436	Congenital anomalies of eyelids, lacrimal system and orbit
				7438	Other specified anomalies of eye
				7439	Unspecified anomalies of eye
				3627	Hereditary retinal dystrophies
				3622	Other proliferative retinopathy
				3743	Ptosis of eyelid
				3787	Other strabisms

GROUPS		SUBGROUPS		CONGENITAL ANOMALIES	
CODE		CODE		ICD CODE	
				3795	Nystagmus and other irregular eye movements
				224	Benign neoplasm of eye
C	Ear	06	Congenital anomalies of ear	7440	Anomalies of ear causing impairment of hearing
				7441	Accessory auricle
				7442	Other specified anomalies of ear
				7443	Unspecified anomalies of ear
D	Cardiovascular system	07	Bulbus cordis anomalies and anomalies of cardiac septal closure	7450	Common truncus
				7451	Transposition of great vessels
				7452	Tetralogy of Fallot
				7453	Common ventricle
				7454	Ventricular septal defect
				7455	Ostium secundum type atrial septal defect
				7456	Endocardial cushion defect
				7457	Cor biloculare
				7458	Other bulbus cordis anomalies and anomalies of cardiac septal closure
				7459	Unspecified defect or septal closure
		08	Other congenital anomalies of heart	7460	Anomalies of pulmonary valve
				7461	Tricuspid atresia and stenosis, congenital
				7462	Ebstein's anomaly
				7463	Congenital stenosis of aortic valve
				7464	Congenital insufficiency of aortic valve
				7465	Congenital mitral stenosis
				7466	Congenital mitral unsufficiency
				7467	Hypoplastic left heart syndrome
				7468	Other specified anomalies of heart
				7469	Unspecified anomalies of heart

GROUPS		SUBGROUPS		CONGENITAL ANOMALIES	
CODE		CODE		ICD CODE	
D	(Cont.)	08	(cont.)	4253	Endocardial fibroelastosis
				7852	Undiagnosed cardiac murmur
		09	Other congenital anomalies of circulatory system	7470	Patent ductus arteriosus
				7471	Coarctation of aorta
				7472	Other anomalies of aorta
				7473	Anomalies of pulmonary artery
				7474	Anomalies of great veins
				7476	Other anomalies of peripheral vascular system
				7478	Other specified anomalies of circulatory system
				7475	Absence or hypoplasia of umbilical artery
				7479	Unspecified anomalies of circulatory system
E	Respiratory system	10	Anomalies of respiratory system	7482	Web of larynx
				7483	Other anomalies of larynx, trachea and bronchus
				7484	Congenital cystic lung
				7485	Agenesis, hypoplasia and dysplasia of lung
				7486	Other anomalies of lung
				7488	Other specified anomalies of respiratory system
				7489	Unspecified anomalies of respiratory system
				2358	Benign neoplasm of pleura, thymus and mediastinum
				7702	Interstitial emphysema and related conditions
				4920	Emphysema

GROUPS		SUBGROUPS		CONGENITAL ANOMALIES	
CODE		CODE		ICD CODE	
F	Cleft palate and/or cleft lip	11	Cleft palate and clekt lip	7490	Cleft palate
				7491	Cleft lip
				7492	Cleft palate with cleft lip
G	Digestive system	12	Tracheo-oesophageal fistula, oesophageal atresia and stenosis	7503	Tracheo-oesophageal fistula, oesophageal atresia and stenosis
		13	Atresia and stenosis of large intestine, rectum and anal canal	75121	Stenosis, atresia or absence of rectum with fistula
				75122	Stenosis, atresia or absence of rectum without mention of fistula
				75123	Stenosis, atresia or absence of anus with fistula
				75124	Stenosis, atresia or absence of anus without mention of fistula
		14	Other anomalies of digestive system and upper alimentary	7504	Other specified anomalies of oesphagus
				7505	Congenital hypertrophic pyloric stenosis
				7507	Other specified anomalies of stomach
				7508	Other specified anomalies of upper alimentary tract
				7509	Unspecified anomalies of upper alimentary tract
				7510	Meckel's diverticulum
				7511	Atresia and stenosis of small intestine
				7513	Hirschsprung's disease and other congenital funtional disorders of colon
				7514	Anomalies of intestinal fixation
				7515	Other anomalies of intestine
				7516	Anomalies of gallbladder, bile ducts and liver

GROUPS		SUBGROUPS		CONGENITAL ANOMALIES	
CODE		CODE		ICD CODE	
				7517	Anomalies of pancreas
				7518	Other specified anomalies of digestive system
				7519	Unspecified anomalies of digestive system
				75120	Atresia and stenosis of large intestine
				211-	Benign neoplasm of digestive system
H	External genitals	15	Anomalies of external genital organs (male)	7525	Undescended testicle
				7528	Other specified anomalies of genital organs
				7786	Congenital hydrocele
				7526	Hypospadias and epispadias
		16	Anomalies of external female genitalia and undeterminate sex	7524	Anomalies of cervix, vagina and external female genitalia
				7527	Indeterminate sex and pseudo-hermaphroditism
I	Internal genitals, urinary tract and kidney	17	Other anomalies of genital organs	7520	Anomalies of ovaries
				7521	Anomalies of fallopian tubes and broad ligaments
				7522	Doubling of uterus
				7523	Other anomalies of uterus
				7529	Unspecified anomalies of genital organs
				219-	Benign neoplasm of uterus
				220-	Benign neoplasm of ovary
				221-	Benign neoplasm of other female genital organs
				222-	Benign neoplasm of male genital organs
		18	Renal anomalies	7530	Renal agenesis and dysgenesis
				7531	Cystic kidney disease
				7533	Other specified anomalies of kidney
				2330	Benign neoplasm of kidney

GROUPS		SUBGROUPS		CONGENITAL ANOMALIES	
CODE		CODE		ICD CODE	
I	(Cont.)	19	~~(cont.)~~ Anomalies of urinary tract	7532	Obstructive defects of renal pelvis ureter
				7534	Other specified anomalies of ureter
				7535	Extrophy of urinary bladder
				7536	Atresia and stenosis of urethra and bladder
				7537	Anomalies of urachus
				7538	Other specified anomalies of bladder and urethra
				7539	Unspecified anomalies of urinary system
				2231	Benign neoplasm of renal pelvis
				2232	Benign neoplasm of ureter
				2233	Benign neoplasm of bladder
				2238	Benign neoplasm of other specified sites
				2239	Benign neoplasm of urinary organs site unspecified
J	Limbs	20	Polydactyly and syndactyly	7550	Polydactyly
				7551	Syndactyly
		21	Reduction deformities of limbs	7552	Reduction deformities of upper limb
				7553	Reduction deformities of lower limb
				7554	Reduction deformities of unspecified limb
		22	Talipes	7545	Varus deformities of feet
				7546	Valgus deformities of feet
				7547	Other deformities of feet
		23	Other anomalies of limbs	7543	Congenital dislocation of hip
				7544	Congenital genu recurvatum and bowing of long bones of leg

GROUPS		SUBGROUPS		CONGENITAL ANOMALIES	
CODE		CODE		ICD CODE	
J	(Cont.)	23	(cont.)	7555	Other anomalies of upper limb including shoulder girdle
				7556	Other anomalies of lower limb including pelvic girdle
				7558	Other specified anomalies of unspecified limb
				7559	Unspecified anomalies of unspecified limb
K	Other anomalies of musculoskeletal and connective tissues	24	Anomalies of tongue, branchial cleft, auricular sinus	7444	Branchial cleft, cyst or fistule; preauricular sinus
				7500	Tongue tie
				7501	Other anomalies of tongue
				7502	Other specified anomalies of mouth and pharynx
		25	Anomalies of nose, face, skull and neck	5240	Major anomalies of jaw size
				5249	Unspecified dentofacial anomalies including malocclusion
				7445	Webbing of neck
				7448	Other specified anomalies of face and neck
				7449	Unspecified anomalies of face and neck
				7480	Choanal atresia
				7481	Other anomalies of nose
				7540	Certain congenital musculoskeletal deformities of skull, face and jaw
				7560	Anomalies of skull and face bones
				5209	Unspecified disorders of tooth development and eruption

GROUPS		SUBGROUPS		CONGENITAL ANOMALIES
CODE		CODE	ICD CODE	
K	(Cont.)	26 Anomalies of diaphragm	7506	Congenital hiatus hernia
			7566	Anomalies of diaphragm
		27 Other musculoskeletal anomalies of thorax and neck	7548	Other specified congenital musculo-skeletal deformities
			7561	Anomalies of spine
			7541	Certain congenital musculoskeletal deformities of sternocleidomastoid muscle
			7542	Certain congenital musculoskeletal deformities of spine
			7562	Cervical rib
			7563	Other anomalies of ribs and sternum
		28 Anomalies of abdominal wall	7567	Anomalies of abdominal wall
			5509	Inguinal hernia without mention of obstruction or gangrene
			5530	Femoral hernia
			5531	Umbilical hernia, omphalocele and para umbilical hernia
			5532	Ventral (incisional) hernia
			5538	Hernia of other specified sites
			5539	Hernia of unspecified site
		29 Other anomalies of bones, muscles, cartilages and connective tissues	7564	Chondrodystrophy
			7565	Osteodystrophies
			7568	Other specified anomalies of muscle, tendon, fascia and connective tissue
			7569	Unspecified anomalies of musculoskeletal system
			2380	Benign neoplasm of bone and articular cartilage

GROUPS		SUBGROUPS		CONGENITAL ANOMALIES
CODE		CODE	ICD CODE	
L	Teguments	30 Anomalies of the integument	7570	Hereditary oedema of legs
			7571	Ichthyiosis congenita
			7572	Dermatoglyphie congenita
			7574	Specified anomalies of hair
			7575	Specified anomalies of nails
			7578	Other specified anomalies of the integument
			7579	Unspecified anomalies of the integument
		31 Skin tags and benign neoplasm of skin	6851	Pilonidal cyst without mention of abscess
			7573	Other specified anomalies of skin
			2140	Lipoma any site
			2169	Benign neoplasm of skin site unspecified
			2280	Haemangioma any site
			2281	Lymphangioma any site
M	Chromosomal	32 Down's syndrome	7580	Down's syndrome
		33 Other chromosomal anomalies	7581	Patau's syndrome
			7582	Edward's syndrome
			7583	Autosomal deletion syndromes
			7584	Balanced autosomal translocation in normal individual
			7585	Other conditions due to autosomal anomalies
			7586	Gonadal dysgenesis
			7587	Klinefelter's syndrome
			7588	Other conditions due to sex chromosome anomalies
			7589	Conditions due to anomaly of unspecified chromosome

GROUPS		SUBGROUPS		CONGENITAL ANOMALIES	
CODE		CODE		ICD CODE	
N	Metabolic defects	34	Metabolic defects	2439	Congenital hypothyroidism
				2461	Dyshormonogenic goitre
				2533	Pituitary dwarfism
				2534	Other anterior pituitary disorders
				2552	Adrenogenital disorders
				2579	Unsepcified testicular hypofunction including pseudohermaphroditism male with gonodal disorder
				2598	Other endocrine disorders
				2700	Disturbances of amino-acid transport
				2701	Phenylketonuria
				2702	Other disturbances of aromatic amino-acid metabolism
				2703	Disturbances of branched-chain amino-acid metablism
				2704	Disturbances of sulphur-bearing amino-acid metabolism
				2705	Disturbances of histidine metabolism
				2706	Disturbances of urea cycle metabolism
				2707	Other disturbances of straight-chain amino-acid metabolism
				2708	Other disorders of amino-acid transport and metabolism
				2709	Unspecified disorders of amino-acid transport and metabolism
				2710	Glycogenosis
				2711	Galactosaemia
				2712	Hereditary fructose intolerance
				2713	Intestinal disaccharidase deficiencies and disaccharide malabsorption
				2718	Other disorders of carbohydrate transport and metabolism

GROUPS		SUBGROUPS		CONGENITAL ANOMALIES	
CODE		CODE		ICD CODE	
N	(Cont.)	34	(cont.)	2719	Unspecified disorders of carbohydrate transport and metabolism
				2720	Pure hypercholesterolaemia
				2721	Pure hyperglyceridaemia
				2722	Mixed hyperlipidaemia
				2723	Hyperchylomicronaemia
				2724	Other and unspecified hyperlipidaemia
				2725	Lipoprotein deficiencies
				2726	Lipodystrophy
				2727	Lipidoses
				2728	Other disorders of lipoid metabolism
				2729	Unspecified disorders of lipoid metabolism
				2730	Polyclonal hypergammaglobulinaemia
				2731	Monoclonal paraprotoinaemia
				2732	Other paraproteinemias
				2733	Macroglobulinaemia
				2738	Other disorders of plasma protein metabolism
				2739	Unspecified, disorders of plasma protein metabolism
				2740	Gouty arthropathy
				2741	Gouty nephropathy
				2748	Gout with other manifestations
				2749	Gout, unspecified
				2750	Disorders of iron metabolism
				2751	Disorders of copper metabolism
				2752	Disorders of magnesium metabolism
				2753	Disorders of phosphorus metabolism
				2754	Disorders of calcium metabolism
				2758	Other disorders of mineral metabolism
				2759	Unspecified disorders of mineral metabolism

GROUPS		SUBGROUPS		CONGENITAL ANOMALIES	
CODE		CODE		ICD CODE	
N	(Cont.)	34	(cont.)	2770	Cystic fibrosis
				2771	Disorders of porphyrin metabolism
				2772	Other disorders of purine and pyrimidine metabolism
				2773	Amyloidosis
				2774	Disorders of bilirubin excretion
				2775	Mucopolysaccharidosis
				2776	Other deficiencies of circulating enzymes
				2778	Other unspecified disorders of metabolism
				2779	Unspecified disorders of metabolism
				2820	Hereditary spherocytosis
				2821	Hereditary elliptocytosis
				2822	Anaemia due to disorders of glutathione metabolism
				2823	Other ihaemolytic anaemia due to enzyme deficiency
				2824	Thalassaemias
				2825	Sickle-cell trait
				2826	Sickle-cell anaemia
				2827	Other haemoglobinopathies
				2860	Congenital factor VIII disorder
				2861	Congenital factor IX disorder
				2862	Congenital factor XI disorder
				2863	Congenital deficiency of other clothing factors
0	Other	35	Anomalies of breast, spleen, adrenal gland and other endocrine glands	7576	Specified anomalies of breast
				7590	Anomalies of spleen
				7591	Anomalies of adrenal gland
				7592	Anomalies of other endocrine glands

GROUPS		SUBGROUPS		CONGENITAL ANOMALIES	
CODE		CODE		ICD CODE	
0	(Cont.)	36	Other syndromes, eponyms and dusease names	2873	Primary thrombocytopenia including thrombocytopenia with absent radius (28732)
				7593	Situs invertus
				7594	Conjoined twins
				7595	Tuberous sclerosis
				7596	Other hamartoses not elsewhere classified
				7598	Other specified anomalies
				7602	Syndromes due to maternal infections including rubella embryopathy
				7710	Congenital rubella
				0900	Congenital syphilis
				7711	Congenital cytoimegalovirus infection
				7712	Other congenital infections
				8888	Syndromes, eponyms and disease names not elsewhere classified
				2879	Unspecified haemorrhagic conditions
				7726	Cutaneous haemorrhage
		37	Other unspecified and ill-defined conditions	7599	Congenital anomaly unspecified
				7597	Multiple congenital anomalies so described
				7998	Other ill-defined conditions
				7799	Unspecified ill-defined conditions originating in the perinatal period
				7780	Hydrops fetalis not due to isoimmunisation
		38	Other		Conditions not elsewhere classified.

APPENDIX VIII : LIST OF DEFECTS SELECTED FOR ROUTINE TABULATION.

BPA CODE	CONGENITAL ANOMALY
740.-	Anencephalus and similar anomalies (including anencephalus, craniorachischisis and iniencephaly)
741.-	Spina bifida with or without mention of hydrocephalus
742.0	Encephalocele
742.1	Microcephalus
742.3	Hydrocephalus
743.0	Anophthalmos
743.1	Microphthalmos
743.32	Congenital cataract
7450	Common truncus
7451	Transposition of great vessels
7452	Tetralogy of Fallot
7453	Common ventricle
7454	Ventricular septal defect
7455	Ostium secundum type atrial septal defect
7456	Ostium primum type atrial septal defect
7470	Patent ductus arteriosus
7471	Coarctation of aorta
7469	Unspecified anomalies of heart
7490	Cleft palate
7491	Cleft lip
7492	Cleft palate with cleft lip
7503	Tracheo-oesophageal fistula, oesophageal atresia and stenosis
7511	Atresia and stenosis of small intestine
7512	Atresia and stenosis of large intestine, rectum and anal canal
7526	Hypospadias and epispadias
7530	Renal agenesis and dysgenesis
7531	Cystic kidney disease
7535	Exstrophy of urinary bladder
7543	Congenital dislocation of hip
7545	Varus deformities of feet
7546	Valgus deformities of feet
75473	Clubfoot NOS
7550	Polydactyly
7551	Syndactyly
7552	Reduction deformities of upper limb
7553	Reduction deformities of lower limb
7554	Reduction deformities, unspecified limb
75661	Anomalies of diaphragm
75670	Exomphalos
75671	Gastroschisis
5531	Omphalocele
7580	Down's Syndrome
7581	Trisomy D (Patau's syndrome)
7582	Trisomy E (Edwards's syndrome)

APPENDIX IX : LIST OF CASES FOR EXCLUSION.

Reports of cases with the following anomalies are not to be transmitted to EUROCAT Central Registry unless occurring in combination with other specified anomalies :

- Spina bifida occulta uncomplicated (756.10).
- Stenosis or stricture of lacrimal duct (743.65).
- Minor or unspecified anomaly of auricle (744.3).
- Minor or unspecified anomaly of nose (748.19).
- Minor or unspecified deformity of face (744.91).
- Minor anomaly of nipple (757.68), accessory or ectopic nipple (757.65).
- Congenital umbilical hernia (553.1), inguinal (550), or para umbilical (553.1).
- Undescended testicle (752.5) and unspecified ectopic testis (752.53).
- Congenital hydrocele or hydrocele of testis (778.6).
- Phymosis (605)
- Hypospadias when the meatus lies before the coronary sulcus (752.60).
- Abnormal palmar crease (757.2).
- Skin tag with surface less than 4 cm^2 : skin tag (757.31), naevus (757.38), angioma (228.0), haemangioma (228.0), glomus tumor (228.0), lymphangioma (228.1), birthmark (757.38).
- Clicking hip (754.32).
- Clubfoot of postural origin (754.73).
- Minor or unspecified anomalies of toe (755.60) such as hallux valgus, hallux varus, or "orteil en marteau".
- Functional or unspecified cardiac murmur (785.2).
- Absence or hypoplasia of umbilical artery, single umbilical artery (747.5).