BIRTH DEFECTS SURVEILLANCE

QUICK REFERENCE HANDBOOK
OF SELECTED CONGENITAL ANOMALIES AND INFECTIONS

World Health Organization

CDC
Centers for Disease Control and Prevention

International Clearinghouse for Birth Defects Surveillance and Research

Birth defects surveillance: quick reference handbook of selected congenital anomalies and infections

ISBN 978-92-4-001541-8 (electronic version)
ISBN 978-92-4-001542-5 (print version)

Design and layout: Green Ink

BIRTH DEFECTS SURVEILLANCE

QUICK REFERENCE HANDBOOK
OF SELECTED CONGENITAL ANOMALIES
AND INFECTIONS

Acknowledgements

The Quick Reference Handbook of Selected Congenital Anomalies is a collaborative effort between the World Health Organization (WHO), the National Center on Birth Defects and Developmental Disabilities (NCBDDD) from the United States Centers for Disease Control and Prevention (CDC), and the International Clearinghouse for Birth Defects Surveillance and Research (ICBDSR).

For their technical input in the preparation of this quick reference handbook, we would like to acknowledge the staff of the NCBDDD, ICBDSR and WHO, particularly the following individuals (in alphabetical order): Mr James K Archer, Dr Jose F Arena, Dr Alejandro Azofeifa, Dr Robert J Berry, Dr Jan Ties Boerma, Dr Lorenzo Botto, Dr Marie Noel Brune Drisse, Ms Grace Davis, Dr Margaret Davis, Dr Luz Maria De-Regil, Dr Pablo Duran, Ms Alissa Eckert, Dr Marcia Feldkamp, Dr Alina Flores, Dr Jaime Frias, Dr Shona Goldsmith, Dr Melba Filimina Gomes, Dr Boris Groisman, Mr Dan J Higgins, Dr Margaret Honein, Ms Jennifer Hulsey, Dr Vijaya Kancherla, Ms Christina Kilgo, Dr Eve Lackritz, Dr Ornella Lincetto, Dr Cara Mai, Dr Elizabeth Mary Mason, Dr Pierpaolo Mastroiacovo, Dr Mario Merialdi, Dr Cynthia Moore, Dr Allisyn Moran, Dr Joseph Mulinare, Dr Teresa Murguia de Sierra, Dr Maria Neira, Dr Richard Olney, Dr Mina Patel, Dr Juan Pablo Pena-Rosas, Dr Nelangi Pinto, Dr Vladimir B Poznyak, Dr Ingrid Rabe, Dr Hilda Razzaghi, Dr Francoise Renaud, Dr Lisa Rogers, Dr Nathalie Roos, Dr Jorge Rosenthal, Dr Csaba Siffel, Dr Haley Smithers-Sheedy, Dr Joseph Sniezek, Dr Gretchen Stevens, Dr Melanie Taylor, Dr Marleen Temmerman, Ms Diana Valencia, Dr Claudia Vellozzi, Dr Severin Von Xylander, and Dr Jennifer Williams.

The drawings were all supplied by CDC/NCBDDD.

We would also like to thank Dr Rajesh Mehta and Dr Neena Raina from the WHO Regional Office for South-East Asia for their valuable feedback during the development process.

We gratefully acknowledge and thank the United States Agency for International Development for providing financial support for this work.

The findings and conclusions in this quick reference handbook are those of the authors and do not necessarily represent the views of the Centers for Disease Control and Prevention.

Abbreviations

AFP	alpha fetoprotein
ASD	atrial septal defect
cCMV	congenital cytomegalovirus
CDC	United States Centers for Disease Control and Prevention
CHARGE	**c**oloboma, **h**eart defects, choanal **a**tresia, growth **r**etardation, **g**enital abnormalities, **e**ar abnormalities
CHD	congenital heart defect
CLIA	chemiluminescence immunoassay
CMV	cytomegalovirus
CNS	central nervous system
CRI	congenital rubella infection
CRS	congenital rubella syndrome
CSF	cerebrospinal fluid
CT	computed tomography
CVS	chorionic villus sampling
CZS	congenital Zika syndrome
DORV	double outlet right ventricle
DQI	data quality indicator
d-TGA	D(dextro)-transposition of the great arteries
ECLAMC	Latin American Collaborative Study of Congenital Malformations
EIA	enzyme immunoassay
ELISA	enzyme-linked immunosorbent assay
ETOP	elective terminations of pregnancy
ETOPFA	elective termination of pregnancy for fetal anomaly
EUROCAT	European Network of Population-Based Registries for the Epidemiological Surveillance of Congenital Anomalies
HC	head circumference
HLHS	hypoplastic left heart syndrome
IAA	interrupted aortic arch

ICBDSR	International Clearinghouse for Birth Defects Surveillance and Research
ICD-9	*International Statistical Classification of Diseases and Related Health Problems, Ninth revision*
ICD-10	*International Statistical Classification of Diseases and Related Health Problems, Tenth revision*
Ig(G/M)	immunoglobulin G/immunoglobulin M
LMIC	low- and middle-income country
MCA	multiple congenital anomalies
MRI	magnetic resonance imaging
MURCS	**mu**llerian, **r**enal, **c**ervicothoracic, **s**omite association
NAATs	nucleic acid amplification tests
NBDPN	National Birth Defects Prevention Network
NCBDDD	National Center on Birth Defects and Developmental Disabilities
NOS	not otherwise specified
NTD	neural tube defect
OAV(S)	**o**culo-**a**uriculo-**v**ertebral (**s**pectrum)
OEIS	**o**mphalocele, **e**xstrophy of the cloaca, **i**mperforate anus, **s**pinal defects
PRNT	plaque reduction neutralization test
RCPCH	Royal College of Paediatrics and Child Health
RENAC	National Network of Congenital Anomalies of Argentina
RPR	rapid plasma regain
RT-PCR	reverse transcriptase polymerase chain reaction
SOP	standard operating procedure
TAR	thrombocytopenia absent radius
TEF (also TOF)	tracheo-oesophageal fistula
TEV	talipes equinovarus
TPHA	Treponema pallidum hemagglutination assay
TPPA	Treponema pallidum particle agglutination assay
USA	United States of America
VACTERL	**v**ertebral, **a**nus, **c**ardiac, **t**rachea, o**e**sophagus, **r**enal, **l**imb
VDRL	Venereal Disease Research Laboratory
WHO	World Health Organization
ZIKV	Zika virus

Contents

CONGENITAL ANOMALIES OF THE NERVOUS SYSTEM: NEURAL TUBE DEFECTS

Neural tube defects (NTDs) affect the brain and spinal cord, and are among the most common of the congenital anomalies (see Fig. 1). *Panel A* shows a cross section of the rostral end of the embryo at approximately three weeks after conception, showing the neural groove in the process of closing, overlying the notochord. The neural folds are the rising margins of the neural tube, topped by the neural crest, and demarcate the neural groove centrally. *Panel B* shows a cross section of the middle portion of the embryo after the neural tube has closed. The neural tube – which will ultimately develop into the spinal cord – is now covered by surface ectoderm (later, the skin). The intervening mesoderm will form the bony spine. The notochord is regressing. *Panel C* shows the developmental and clinical features of the main types of NTDs. The diagram in the centre is a dorsal view of a developing embryo, showing a neural tube that is closed in the centre but still open at the cranial and caudal ends. The dotted lines marked A and B refer to the cross sections shown in *panels A* and *B*. Shaded bars point to the region of the neural tube relevant to each defect.

The most prevalent types of NTDs are anencephaly, encephalocele and spina bifida. In anencephaly, the absence of the brain and calvaria can be total or partial. Craniorachischisis is characterized by anencephaly accompanied by a contiguous bony defect of the spine and exposure of neural tissue. In open spina bifida, a bony defect of the posterior vertebral arches (in this case, the lower thoracic vertebrae) is accompanied by herniation of neural tissue and meninges and is not covered by skin. In iniencephaly, dysraphia in the occipital region is accompanied by severe retroflexion of the neck and trunk. In encephalocele, the brain and meninges herniate through a defect in the calvaria. In closed spina bifida, unlike open spina bifida, the bony defect of the posterior vertebral arches (in this case, the lumbar vertebrae), the herniated meninges and neural tissue are covered by skin.

Fig. 1. Neural tube defects