Eurocat

European Surveillance of Congenital Anomalies

Short EUROCAT Report:

Congenital Anomalies Diagnosed After Age 1 Year and Their Impact on Prevalence

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Background

At the PMC meeting in 2007 the Steering Committee discussed an application from a new registry that included new cases up to the age of 18 years. This led to a discussion of the impact of late diagnosed cases on overall prevalence of congenital anomalies and prevalence in specific subgroups (see EUROCAT website for subgroup definitions).

Method

Ruth and Ester have looked at the data in the Central Database in order to describe the impact of cases diagnosed after age 1 year. Years 2000-2005, cases diagnosed after one year of age: code 5 in the "when discovered" variable were analysed.

Results

Seven out of 36 registries had more than 2 % of their cases diagnosed after one year (Table 1 and Figure). These registries are Odense, Northern Netherlands, Strasbourg, Vaud, Styria, Sicily and Wales. Fifteen registries did not report any cases diagnosed after one year and the remaining registries had less than one percent of cases diagnosed after one year.
The impact of late diagnosed cases on the subgroups is presented in Table 2 including the seven registries with 2-5% late diagnosed cases. More than 10% of the cases where diagnosed after one year in the subgroups microcephaly, eye, teratogenic syndromes and fetal alcohol syndrome. Subgroups with 5-10% late diagnosed cases were nervous system, all specific eye subgroups except anophthalmos, coarctation of aorta, Hirschprung, posterior urethral valves, craniosynostosis, genetic syndromes and microdeletions, Klinefelter syndrome and Cri-du-chat syndrome.

Conclusion

In general less than five percent of all congenital malformations are diagnosed after one year of age. For some subgroups a rather large proportion of cases are diagnosed after one year. This has to be taken into account if specific studies on these subgroups are performed.

Ruth Greenlees and Ester Garne
December 2008

Table 1

Registry	Total cases	Number diagnosed >1 year	% diagnosed >1year
Hainaut	1983	0	0.00
Odense	827	24	2.90
Paris	7707	0	0.00
Tuscany	3469	1	0.03
Dublin	2702	6	0.22
N Netherlands	2384	63	2.64
Emilia Romagna	3481	0	0.00
Strasbourg	1733	43	2.48
Vaud	1626	42	2.58
Zagreb	537	4	0.74
Malta	734	1	0.14
NE Italy	4085	0	0.00
S Portugal	1068	0	0.00
Antwerp	2770	0	0.00
Basque Country	2153	9	0.42
Saxony Anhalt	3514	6	0.17
Mainz	1051	0	0.00
Barcelona	982	0	0.00
Styria	2160	62	2.87
N Thames	4005	2	0.05
Cork and Kerry	934	6	0.64
Sicily	1263	35	2.77
Campania	3383	0	0.00
Wales	7137	338	4.74
Norway	13342	0	0.00
Auvergne	374	0	0.00
Ukraine	558	0	0.00
Reunion	1601	1	0.06
Wielkopolska	4554	11	0.24
Thames Valley	1172	0	0.00
Wessex	2997	4	0.13
Trent	7905	4	0.05
Northern Region	4317	0	0.00
Hungary	8154	0	0.00
Poland	21308	36	0.17
SE Ireland	681	3	0.44
Overall	**128651**	**701**	**0.54**

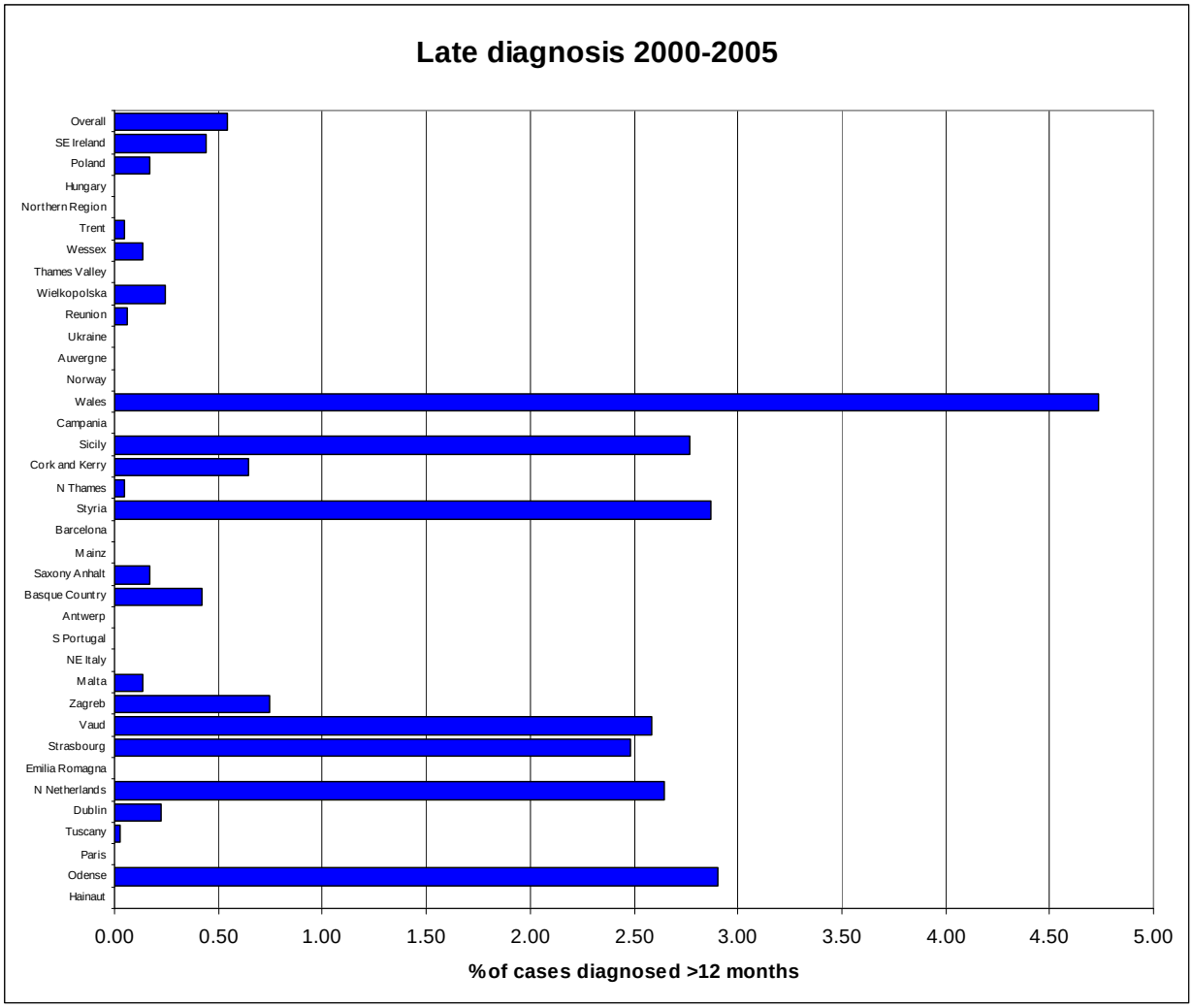
Late diagnosis 2000-2005
Overall
SE Ireland
Poland
Hungary
Northern Region
Trent
Wessex
Thames Valley
Wielkopolska
Reunion
Ukraine
Auvergne
Norway
Wales
Campania
Sicily
Cork and Kerry
N Thames
Styria
Barcelona
Mainz
Saxony Anhalt
Basque Country
Antwerp
S Portugal
NE Italy
Malta
Zagreb
Vaud
Strasbourg
Emilia Romagna
N Netherlands
Dublin
Tuscany
Paris
Odense
Hainaut
0.00
0.50
1.00
1.50
2.00
2.50
3.00
3.50
4.00
4.50
5.00
% of cases diagnosed >12 months

Table 2
Cases diagnosed after age one year by subgroup from Odense, Northern Netherlands, Strasbourg, Vaud, Styria, Sicily and Wales

		Number late diagnosed	Total LBs	% late diagnosed
al1	All anomalies	607	14747	4.1
al2	Nervous system	43	694	6.2
al3	Neural Tube Defects	1	152	0.7
al4	Anencephalus and similar	0	15	0.0
al5	Encephalocele	0	21	0.0
al6	Spina Bifida	1	116	0.9
al7	Hydrocephaly	6	172	3.5
al8	Microcephaly	19	153	12.4
al9	Arhinencephaly/ holoprosencephaly	0	23	0.0
al10	Eye	47	419	11.2
al11	Anophthalmos/ micropthalmos	5	75	6.7
al12	Anophthalmos	0	13	0.0
al13	Congenital cataract	10	116	8.6
al14	Congenital glaucoma	2	22	9.1
al15	Ear, face and neck	7	209	3.3
al16	Anotia	0	8	0.0
al17	Congenital heart disease	180	5198	3.5
al18	Common arterial truncus	4	101	4.0
al19	Transposition of great vessels	2	174	1.1
al20	Single ventricle	0	27	0.0
al21	Ventricular septal defect (VSD)	51	2086	2.4
al22	Atrial septal defect (ASD)	60	2061	2.9
al23	Atrioventricular septal defect (AVSD)	2	186	1.1
al24	Tetralogy of Fallot	1	151	0.7
al25	Tricuspid atresia and stenosis	0	34	0.0
al26	Ebstein's anomaly	1	23	4.3
al27	Pulmonary valve stenosis	17	428	4.0
al28	Pulmonary valve atresia	0	47	0.0
al29	Aortic valve atresia/stenosis	5	105	4.8
al30	Hypoplastic left heart	1	90	1.1
al31	Hypoplastic right heart	0	16	0.0
al32	Coarctation of aorta	15	258	5.8
al33	Total anomalous pulm venous return	0	45	0.0
al34	Respiratory	10	394	2.5
al35	Choanal atresia	1	83	1.2
al36	Cystic adenomatous malf of lung	1	26	3.8
al37	Oro-facial clefts	14	893	1.6
al38	Cleft lip with or without palate	0	517	0.0
al39	Cleft palate	14	376	3.7
al40	Digestive system	22	1032	2.1
al41	Oesophageal atresia	0	128	0.0
al42	Duodenal atresia or stenosis	0	46	0.0
al43	Atresia or stenosis of other parts of small intestine	0	56	0.0
al44	Ano-rectal atresia and stenosis	3	167	1.8
al45	Hirschsprung's disease	7	100	7.0
al46	Atresia of bile ducts	0	37	0.0
al47	Annular pancreas	0	13	0.0

		Number late diagnosed	LBs only	% late diagnosed
al48	Diaphragmatic hernia	4	129	3.1
al49	Abdominal wall defects	0	230	0.0
al50	Gastroschisis	0	148	0.0
al51	Omphalocele	0	74	0.0
al52	Urinary	62	2025	3.1
al53	*Bilateral* renal agenesis including Potter syndrome	0	24	0.0
al54	Cystic kidney disease	0	256	0.0
al55	Congenital hydronephrosis	17	1073	1.6
al56	Bladder extrophy and/or epispadia	0	37	0.0
al57	Posterior urethral valve and/or prune belly	4	63	6.3
al58	Genital	20	1464	1.4
al59	Hypospadias	11	1199	0.9
al60	Indeterminate sex	1	30	3.3
al61	Limb	54	2388	2.3
al62	Limb reduction	2	287	0.7
al63	Upper limb reduction	0	213	0.0
al64	Lower limb reduction	0	87	0.0
al65	Complete absence of a limb	0	1	0.0
al66	Club foot - talipes equinovarus	3	487	0.6
al67	Hip dislocation and/or dysplasia	30	739	4.1
al68	Polydactyly	0	464	0.0
al69	Syndactyly	2	317	0.6
al70	Arthrogryposis multiplex congenita	1	35	2.9
al71	Musculo-skeletal	26	610	4.3
al72	Thanatophoric dwarfism	0	3	0.0
al73	Jeunes syndrome	0	3	0.0
al74	Achondroplasia	0	17	0.0
al75	Craniosynostosis	13	189	6.9
al76	Congenital constriction bands/amniotic band	1	27	3.7
al77	Other malformations	47	543	8.7
al78	Asplenia	0	7	0.0
al79	Situs inversus	0	30	0.0
al80	Conjoined twins	0	0	-
al81	Disorders of skin	14	348	4.0
al82	Teratogenic syndromes with malformations	5	48	10.4
al83	Fetal alcohol syndrome	3	19	15.8
al84	Valproate syndrome	0	4	0.0
al85	Warfarin syndrome	0	1	0.0
al86	Maternal infections resulting in malformations	1	22	4.5
al87	Genetic syndromes & microdeletions	24	411	5.8
al88	Chromosomal	36	874	4.1
al89	Down's syndrome	4	472	0.8
al90	Patau syndrome/ trisomy 13	1	31	3.2
al91	Edward syndrome/ trisomy 18	0	47	0.0
al92	Turner's syndrome	1	48	2.1
al93	Klinefelter's syndrome	3	37	8.1
al94	Cri-du-chat syndrome	1	12	8.3
al95	Wolff-Hirschorn syndrome	0	8	0.0