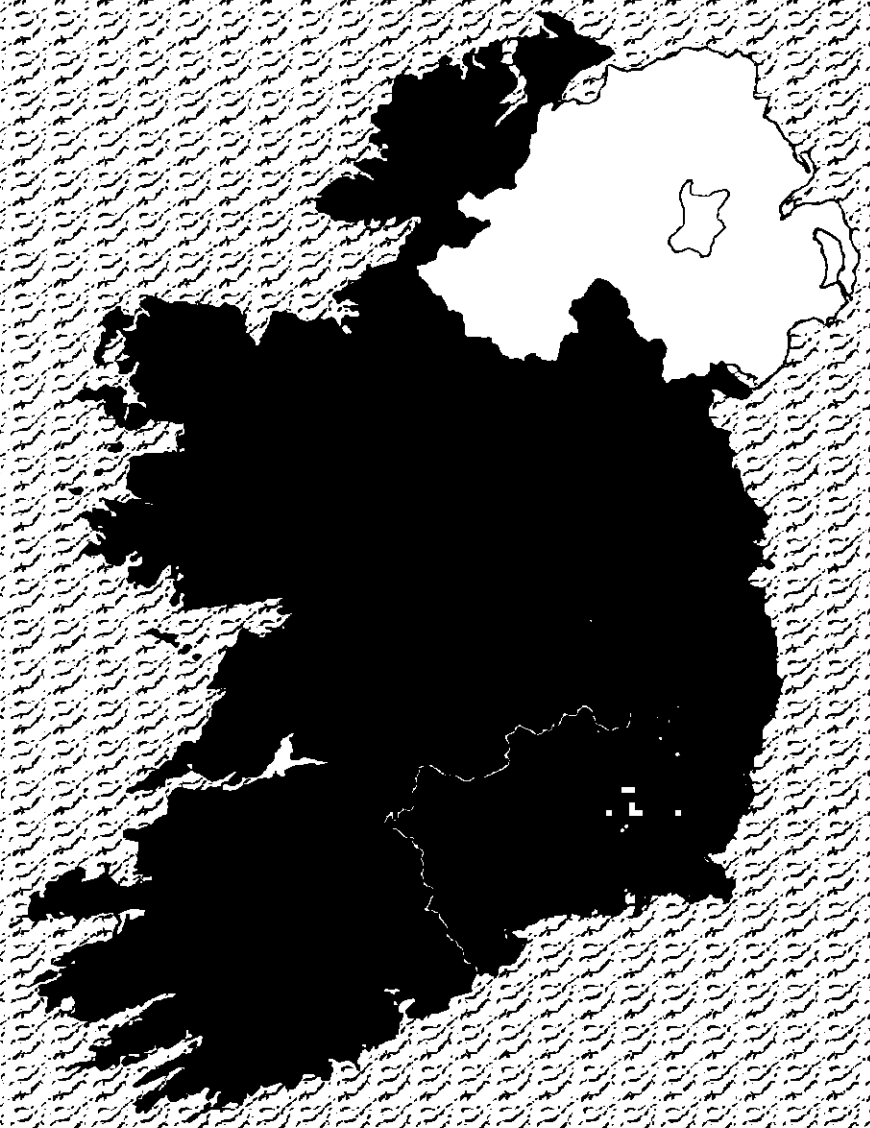
...ies in the East of Ireland
...97 - 2001



Report of the Birth Defect Registries of the Eastern Regional Health Authority North Eastern Health Board South Eastern Health Board

2004

Feidhmeannacht na Seirbhíse Sláinte
Health Service Executive

With Compliments

Dublin EUROCAT Registry of Congenital Anomalies

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EXECUTIVE SUMMARY

Congenital anomalies are an important cause of peri-natal mortality. In addition, major anomalies such as neural tube defects (NTD) and chromosomal anomalies have lifelong consequences for individuals themselves, their families and the health and social services.

Surveillance of congenital anomalies is an important activity in any health service as a means of monitoring trends in birth defects, early detection of unexpected rises in specific birth defects, and in evaluating the outcome of health interventions and research. To date, there has been a paucity of information on congenital anomalies in Ireland, mainly due to the lack of congenital anomaly surveillance for much of the country. Until 1996, surveillance of birth defects by health board population based registries was undertaken in four of the twenty six counties of the Republic of Ireland - counties Dublin, Wicklow and Kildare (Dublin EUROCAT Register of the Eastern Regional Health Authority) and in county Galway (Galway EUROCAT Register). Some surveillance of congenital anomalies was also being undertaken in other areas. In 1997, three new registries were established, in the Southern Health Board, the South-Eastern Health Board (SEHB) and the North Eastern Health Board (NEHB). Because of the geographical proximity of the latter two health boards with the Eastern Regional Health Authority (ERHA), a co-operative approach was adopted for surveillance along the eastern seaboard of Ireland. Between them, these three regions have a catchment of more than half of the births in the country. This report is the first publication of data from five years of surveillance of congenital anomalies in the three regions (East of Ireland) from 1997-2001 and what follows is a brief outline of what is contained in the report.

- There were 3,842 children born with congenital anomalies in the East of Ireland from 1997-2001, representing 2.4% of all births during the five-year period. As the highest concentration of births occurred in the Dublin area, the trend in congenital anomalies in the East as a whole was determined by that in the catchment area of the Eastern Regional Health Authority.

- Although the birth prevalence rate of NTD remained relatively stable during the five-year period (following a large decline during the 1980s and early 1990s), there were nevertheless 164 children born with a NTD. It is known that more than 50% of NTD can be prevented through adequate dietary intake of the vitamin folic acid.

- With regard to chromosomal anomalies, a gradual rise in the number and rate of Trisomy 21 (Down syndrome), Trisomy 13 (Patau's syndrome) and Trisomy 18 (Edward's syndrome) occurred during the five years. This is primarily as a result of increasing maternal age in recent years and is a phenomenon that has been observed elsewhere in Europe. The increase has not been uniform, showing peaks in 1998 and 2000. Holoprosencephaly may also be associated with chromosomal trisomies and a concomitant rise was also observed during the period.

- There were 958 children born with a congenital heart defect during the five years, most of whom would require surgical or other specialist treatment. In 1999, more cases of hypoplastic left heart, transposition of the great vessels and coarctation of the aorta were observed, the reasons for which are unclear.

- The birth prevalence rate for facial cleft anomalies fluctuated from year to year, although it was not within the scope of this report to examine in more detail the individual types of facial cleft, which may have shown more pronounced trends.

- There were 119 children born with a major digestive tract anomaly such as tracheo-oesophageal fistula, intestinal atresia/stenosis or ano-rectal atresia/stenosis, anomalies that usually require early surgery. The overall rate of each was stable from year to year.

- Many minor limb anomalies tend to be ascertained later than major limb defects. This was evident with a decrease in 2001 in the birth prevalence of all limb anomalies combined. A higher rate of polydactyly was observed in recent years and particularly in 2001 with the influx of African immigrants, among whom a number of families had members with polydactyly, it being frequently a genetically dominant inherited trait.

- With regard to abdominal wall anomalies, an increasing trend was evident for both gastroschisis (1997 to 2000) and omphalocoele (1997 to 2001). The rise in gastroschisis in the East of Ireland mirrors a similar unexpected rise in recent years in other countries in Europe, mostly among children born to younger mothers. The rise in the number of children born with an omphalocoele has accompanied that of chromosomal anomalies, with there being a known association with specific chromosomal trisomies.

- The rates of renal anomalies fluctuated during the period. Renal agenesis and polycystic kidney disease are inter-related, in that the case definition has changed in the past decade with new technology. Some cases diagnosed as renal agenesis formerly, are known to have small cysts and are therefore polycystic kidneys, although the overall number of these anomalies would be the same.

- The rate of external genital anomalies remained stable throughout the period.

- The rate for eye anomalies showed a peak in 1998 and 1999.

- Cystic fibrosis is frequently ascertained late as it is usually not diagnosed until after birth. As a result, the numbers in 2000 and 2001 are less than previous years.

This report is published at a time of major structural change within the Irish health service as a consequence of the abolition of the previous health board structures. Such changes are at an administrative and service provision level, while the burden of disease remains the same regardless of administrative boundaries. Currently, there is no congenital anomaly surveillance for almost one third of births in the country, where no registries are in place. This is an unfortunate deficit for the health service of a modern country like Ireland, unlike other European countries such as Norway, Finland, England, Wales and Sweden, which have national congenital anomaly surveillance systems. Congenital anomaly surveillance is relatively inexpensive to undertake and maintain. It is also much more cost-effective and timely than initiating ad-hoc surveys to investigate a rise in a specific birth defect in a particular region, where there is no such surveillance. The extension of congenital anomaly surveillance nationally would be a prudent step in the context and framework of a progressive health service.

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INTRODUCTION

Birth defects occur in approximately 2.5% of births. The cause of many is frequently unknown although a number of serious birth defects have been shown to be clearly associated with specific risks. For example, neural tube defects are known to be associated with low folic acid intake by prospective mothers prior to conception and in early pregnancy; also, there is an increased risk of giving birth to a child with a chromosomal anomaly with advancing maternal age. In recent years, a slightly increased risk of specific birth defects has been demonstrated to be associated with residence in proximity to hazardous waste landfill dumps. In addition, a steep rise in the occurrence of the abdominal wall defect, gastroschisis, has been thought to be influenced by a variety of factors including cigarette smoking, poor nutrition and possibly illicit drug intake.

As there are more than 60,000 births per year in the Republic of Ireland, one would expect 1,500 children to be born with a birth defect each year. It is not possible to confirm this, as surveillance of birth defects in the country is limited. The actual number could be lower or even higher. One long established surveillance system, has been providing surveillance of birth defects in the east of the country (counties Dublin, Wicklow and Kildare) since 1980, this is the congenital anomaly registry of the Eastern Regional Health Authority (ERHA). The registry is a member of EUROCAT[1,2,3], a European network of population-based registers for the surveillance of congenital anomalies in regions of Europe, and its international counterpart, the International Clearinghouse for Birth Defect Monitoring Systems (ICBD). The only other formal registry in the country until 1997 was in Galway, providing surveillance of birth defects for County Galway. There was some congenital anomaly surveillance in other parts of the country. In 1997, birth defect registries were established in three additional health boards, the North Eastern Health Board (NEHB - counties Louth, Meath, Cavan, Monaghan), the South Eastern Health Board (SEHB - counties Wexford, Waterford, Carlow, Kilkenny, Tipperary South) and the Southern Health Board (SHB - counties Cork, Kerry). Unfortunately, due to many factors, the NEHB had to temporarily suspend the births defect surveillance system at the end of 2001. In addition, there is currently no surveillance of birth defects in the remainder of the country, including the mid-west, the north-west, the midlands and the west outside of Galway. Nor is there any national co-ordination of birth defect surveillance to give a national 'picture', as exists in a number of European countries.

Due to the geographical proximity of the birth defect surveillance systems in the east of the country, the registries of the ERHA, SEHB and NEHB joined in collaborative surveillance of the entire eastern seaboard of the Republic of Ireland (Appendix 1). As the birth defect registries of the NEHB and SEHB are too small to become members of these networks in their own right, it was considered that they would come under the 'umbrella' of the Eastern Region's congenital anomaly surveillance system for linkage with the European and international networks. This report represents the findings of the three surveillance systems in the three health board/authority regions (East of Ireland) for the five years from 1997-2001.

OBJECTIVES OF BIRTH DEFECT REGISTRIES

The specific objectives of birth defect registries are:

(i) Provide baseline epidemiological information on congenital anomalies in the catchments of the ERHA, NEHB and SEHB;

(ii) Detect and investigate trends in the frequency of congenital anomalies in order to assess the impact of known or suspected risk factors;

(iii) Evaluate the effectiveness and efficiency of health services (primary prevention and treatment);

(iv) Provide a well-documented database for etiologic and clinical research;

(v) Act as an information centre that can respond to specific needs, such as the assessment of the impact of environmental accidents or change, or the suspicion of teratogenic influences from food, drugs or other exposures.

METHODOLOGY

Birth defects are defined as structural defects (congenital malformations, deformations, disruptions and dysplasias), chromosomal abnormalities, inborn errors of metabolism, and hereditary diseases. Birth defect registries in the EUROCAT network use the common nomenclature and code system of the British Paediatric Association (BPA) Classification of Diseases[4]. This is a 5-digit code and an extension of the 9th revision of the International Classification of Diseases (ICD 9)[5]. Up to eight congenital anomalies may be coded for each baby. In addition, if a syndrome is recognised it may also be coded. In 1993, a further 6-digit extension that allows for more detailed description of the anomalies was introduced. From 2001, all anomalies are coded according to the 10th revision of the International Classification of Diseases (ICD 10). Codes used for each anomaly and group are available in Appendix 2.

The subject population of the three registries is comprised of all babies born to mothers resident at birth in the counties of Dublin, Wicklow and Kildare (ERHA catchment), Louth, Meath, Monaghan, Cavan (NEHB) and Wexford, Kilkenny, Carlow, Waterford and Tipperary South (SEHB) from 1997-2001. This coverage accounts for approximately 58% of all births in the state. Data are collected from birth to five years of age as some anomalies may be obvious at birth such as spina bifida, with others not becoming apparent until early childhood, for example, some forms of congenital heart disease.

Information is collected on the mother's obstetric record. Data on the length of gestation, type of birth, birth weight, outcome and malformations present are collected as are data on frequency of antenatal diagnosis and results of karyotyping and post-mortem examinations. The registries operate an active surveillance system. Cases are ascertained using multiple sources which include: birth notifications, data from the Hospital Inpatient Enquiry System (HIPE), death certificates, pathology reports, long term illness and records of recipients of the Domiciliary Care Allowance. Other sources within individual hospitals are also used, such as cardiology outpatient letters, admissions to the Special Care Baby Unit of one of the Dublin maternity hospitals, voluntary support groups and the community care services (Senior Area Medical Officers).

Researchers with qualification and experience in nursing or paediatric nursing with training in coding provided by EUROCAT network collect the data on a standardised form from these sources. The anonymised data is entered on specialised software developed by EUROCAT. Access to the registry data is restricted to a nominated account, protected by password.

Data are analysed locally and also at the central EUROCAT registry in the University of Ulster in Northern Ireland. Birth denominator data and maternal birth characteristics data are supplied by the Central Statistics Office. Low birth weight is defined as a birthweight under 2500 grams and premature births are those where the length of gestation is less than 37 weeks. Stillbirth is defined as stillborn weighing 500 grams or more, or at gestational age of 24 weeks or more.

Annual birth prevalence rates for the major anomalies are calculated within registries, and anonymised data are transmitted to the EUROCAT central registry for comparison with other registries. Local data analysis provides:

- prevalence rates by year of birth for all congenital anomalies
- prevalence rates by year of birth for selected anomalies
- descriptive statistics for selected anomalies (male/female ratio, proportion stillborn, mean maternal age etc.)

In addition, research has been carried out in the past by the ERHA registry relating to specific congenital anomalies. This registry also has responded to local concerns regarding increased frequencies of congenital anomalies in geographically defined areas.

The staff of the registries (Appendix 3) are generally part-time and include a Specialist in Public Health Medicine, a research nurse and some secretarial support.

BIRTH CHARACTERISTICS OF THE MATERNAL POPULATION

Based on 2003 data, approximately 58% (35,592 / 61,517) of births (excluding stillbirths) in the country are to residents in the three regions in the east of the country, the largest being the catchment of the ERHA. This percentage remains fairly constant from year to year. In the study period 1997-2001 there were 157,439 live births to women resident in the three regions combined. The number of births increased gradually from 30,104 in 1997 to 33,637 in 2001, an overall rise of 11.7% (Table 1 and Appendix 4).

Maternal age

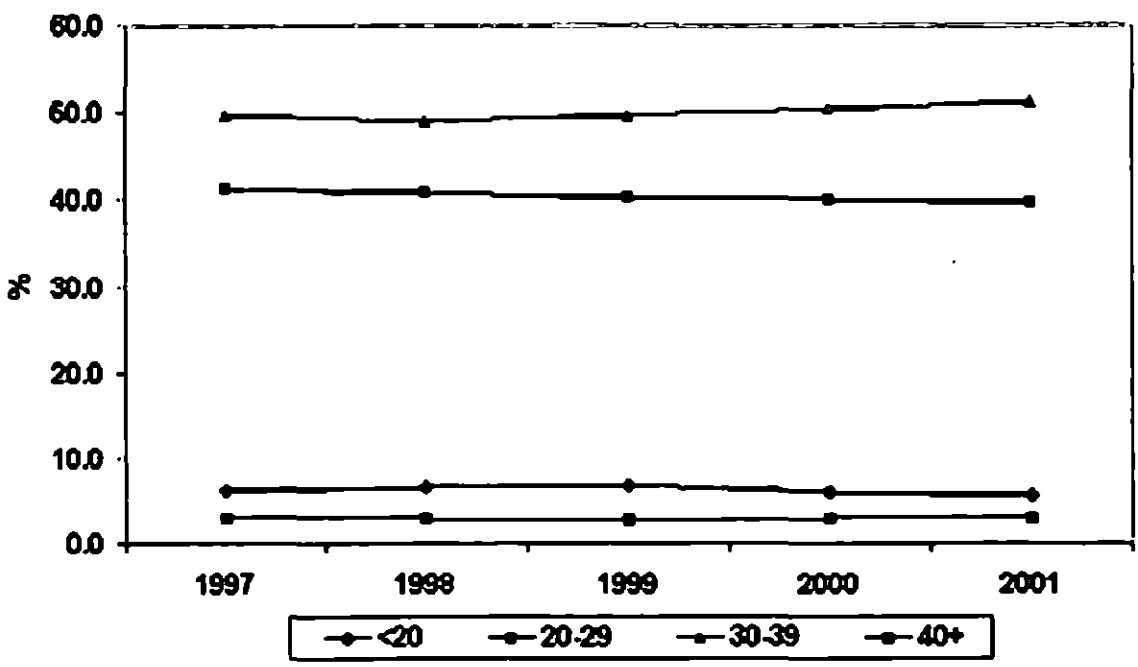
In terms of maternal age, a rise was evident in the numbers of births to most age groups from 1997-2001, particularly in the 30-34 and 35-39 year age groups.

Table 1. Births by maternal age in the East of Ireland - 1997-2001

Age group	1997	1998	1999	2000	2001	Total
15-19	1,846	2,055	2,107	1,919	1,909	9,836
20-24	4,376	4,586	4,604	4,730	5,009	23,305
25-29	8,046	8,052	7,960	7,944	8,338	40,340
30-34	10,114	10,039	10,201	10,354	11,227	51,935
35-39	4,813	5,096	5,273	5,639	5,994	26,815
40-44	830	878	844	919	996	4,467
45+	42	32	32	38	35	179
Unknown	37	145	121	130	129	562
Total	**30,104**	**30,883**	**31,142**	**31,673**	**33,637**	**157,439**

Overall, the proportion of births to women in their thirties increased to over 50% of total births during the period (Fig. 1), with a corresponding slight fall in the proportion of births to women in their twenties to less than 40%. The proportion of births to women in their teens and their forties remained fairly constant.

Fig. 1 Percentage of births by 10-year maternal age group in the east from 1997-2001



Marital status

The marital status of mothers is shown in Table 2. During the total period, two thirds of mothers were married and slightly under one-third were single mothers, with slight variation between health boards /authority.

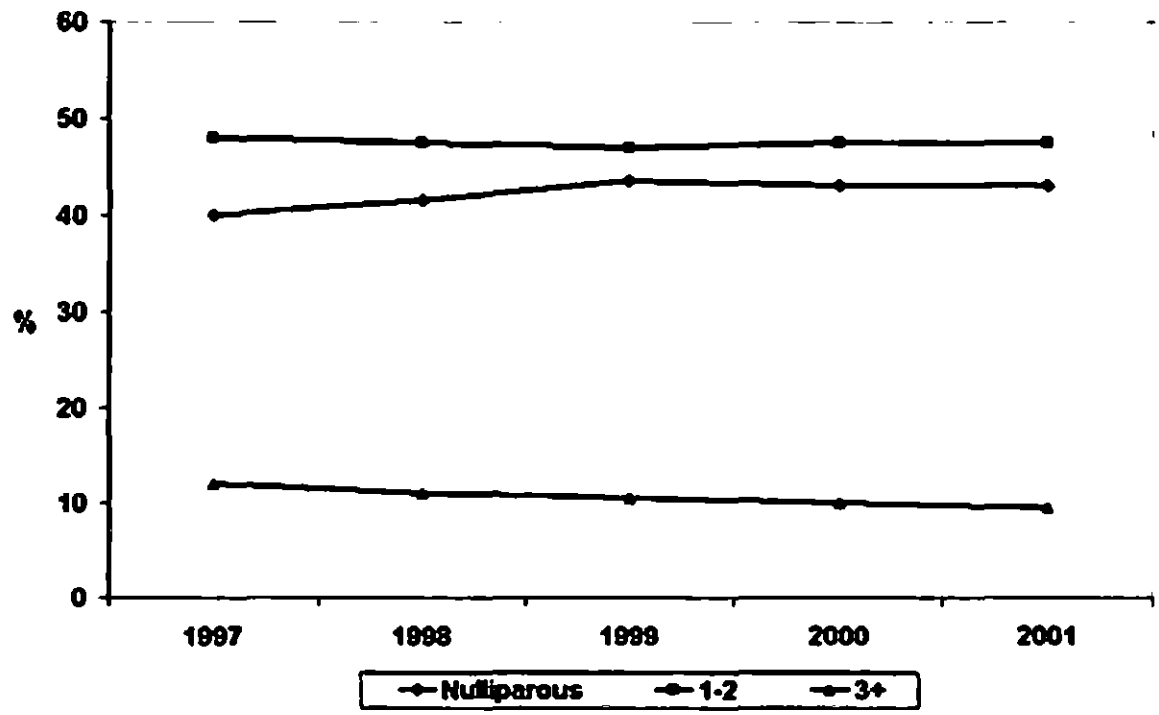
Table 2. Percentage of births by marital status in the East of Ireland 1997-2001

Marital status	EAST	ERHA	SEHB	NEHB
Married	**66.5**	64.5	68.2	73.1
Single	**31.2**	33.0	29.6	24.8
Widowed	**0.1**	0.1	0.1	0.2
Separated	**1.9**	2.0	1.9	1.6
Divorced	**0.3**	0.3	0.2	0.2
Total	**100.0**	100.0	100.0	100.0

Parity

Trends in the number of children previously born (both live births and still births) to mothers in the East of Ireland are shown in Fig. 2. The percentage of children born to mothers with one or two previous births remained stable during the five-year period, while the percentage born to nulliparous mothers rose steadily from 40% in 1997 to 43% in 2001. A fall occurred in the percentage of children born to mothers who had three or more previous births during the same period, from 12% in 1997 to less than 10% in 2001. These patterns reflect a trend evident throughout the 1980s and 1990s towards smaller average family size and increased proportion of births to single women.

Fig. 2. Parity in the East of Ireland from 1997-2001



Stillbirths

The numbers of stillbirths and the stillbirth rate per 1,000 births from 1997-2001 are shown in Table 3. The number of stillbirths showed a slight fall from 1997-1999 and increased thereafter. The overall rate per 1,000 births was 6.1 and varied little from year to year, ranging from 6.5 per 1,000 births in 1997 to 5.8 in 1999.

Table 3. Numbers of livebirths and stillbirths in the East of Ireland from 1997-2001 with the stillbirth rate per 1,000 births

	EAST				ERHA		SEHB		NEHB	
YEAR	LB	SB	TOTB	SB RATE	LB	SB	LB	SB	LB	SB
1997	30,104	196	30,300	6.5	20,156	129	5,450	33	4,498	34
1998	30,883	186	31,069	6.0	20,781	118	5,596	34	4,506	34
1999	31,142	183	31,325	5.8	20,641	128	5,743	28	4,758	27
2000	31,673	195	31,868	6.1	21,097	139	5,779	27	4,797	29
2001	33,637	213	33,850	6.3	22,013	139	6,279	41	5,345	33
Total	157,439	973	158,412	6.1	104,688	653	28,847	163	23,904	157

Note: LB=livebirths, SB=stillbirths, TOTB=LB+SB

DESCRIPTIVE FORMAT OF THE REPORT

Congenital anomalies recorded during the period 1997-2001 are described on the following pages. Each section is devoted to a congenital anomaly category with the number of cases and birth prevalence rate per 10,000 total births (i.e. livebirths and stillbirths) in the three individual health boards/authority and the East of Ireland as a whole shown in tabular form.

Ascertainment of major anomalies and most less serious anomalies is usually complete within a year of birth, whereas some less serious anomalies may not be apparent until more than a year after birth. In addition, with the development of the newer birth defect surveillance systems in the NEHB and SEHB, ascertainment took a longer period of time than in the long-established ERHA registry. Thus, although the total number of anomalies for 2001 as indicated in the next chapter may be slightly underestimated, the corresponding data on specific major anomalies for 2001 in the subsequent sections is mostly complete.

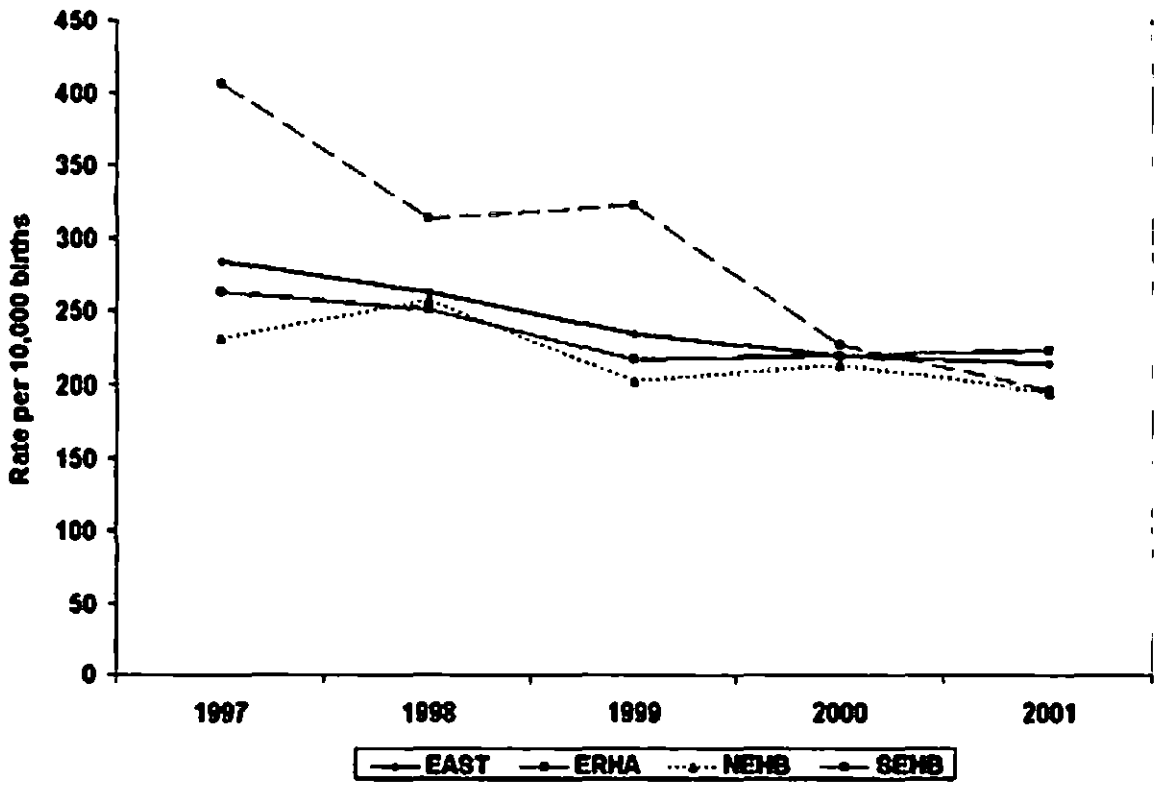
ALL ANOMALIES

There are 700-800 children born in the East of Ireland each year with birth defects. These birth defects range from major lethal anomalies such as some types of neural tube defects or chromosomal anomalies to less serious anomalies such as talipes and polydactyly. The apparent decline in the numbers of defects in 2000 and 2001 is likely to be a result of under-ascertainment in these years rather than a real decline, as a proportion of birth defects are not diagnosed or ascertained until some years after the child is born. (Table 4, Fig. 3).

Table 4. Numbers and rate per 10,000 births: all anomalies in the East of Ireland 1997-2001

	EAST			ERHA			NEHB			SEHB		
YEAR	Births	No.	Rate /10,000	Births	No.	Rate 10,000	Births	No.	Rate /10,000	Births	No.	Rate /10,000
1997	30,300	862	284.5	20,285	534	263.2	4,532	105	231.7	5,483	223	406.7
1998	31,092	819	263.4	20,899	525	251.2	4,540	117	257.7	5,653	177	313.1
1999	31,324	734	234.3	20,767	451	217.2	4,786	97	202.7	5,771	186	322.3
2000	31,861	702	220.3	21,229	467	220.0	4,826	103	213.4	5,806	132	227.4
2001	33,835	725	214.3	22,137	496	224.1	5,378	105	195.2	6,320	124	196.2
Total	158,412	3842	242.5	105,317	2473	234.8	24,062	527	219.0	29,033	842	290.0

Fig. 3 All anomalies: trend in birth prevalence rates per 10,000 births 1997-2001



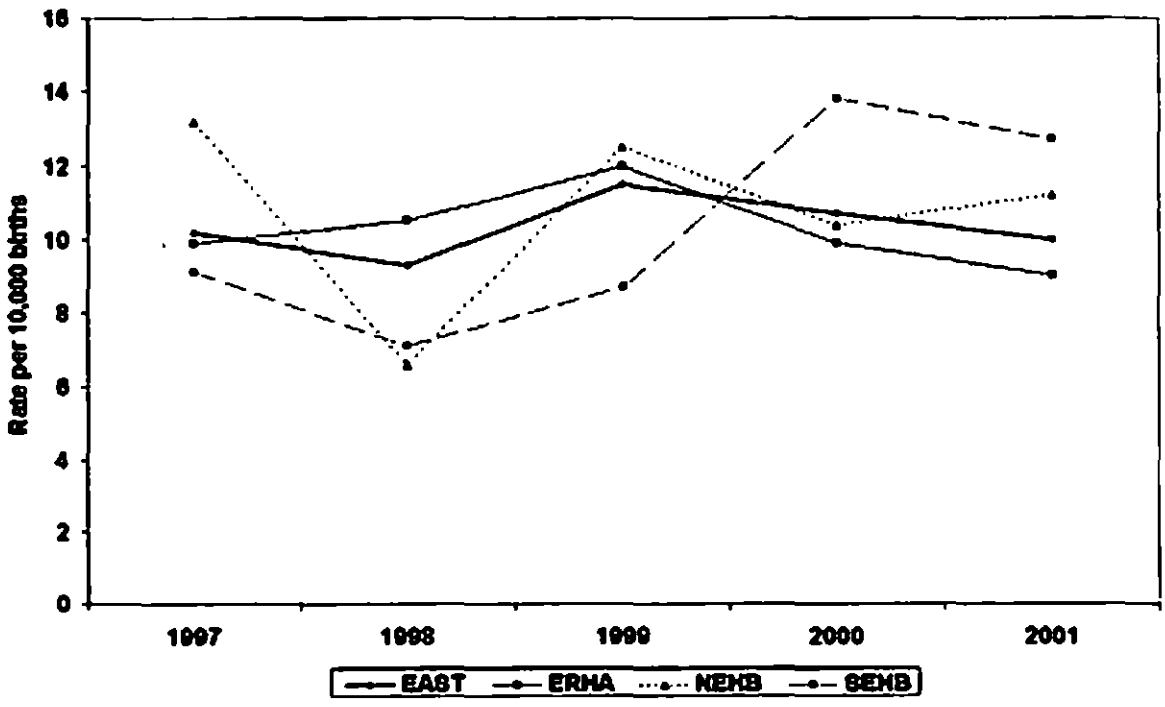
NEURAL TUBE DEFECTS

Neural tube defects (NTD) are the largest group of anomalies of the central nervous system and are comprised of spina bifida, anencephaly, and encephalocoele. Spina bifida and anencephaly encompass the vast majority of NTD. Spina bifida is an anomaly of the spinal cord and is usually associated with severe lifelong disability; anencephaly is a failure of development of the brain and is therefore a lethal anomaly that is incompatible with survival; encephalocoele is a congenital malformation characterised by herniation of the brain and/or meninges through a defect in the skull. At least 50% of NTD is preventable if prospective mothers take the vitamin folic acid in the periconceptional period. There are no specific demographic characteristics of mothers or children with NTD, other than a slight preponderance of female births attributable mainly to anencephaly. During the five-year period, 164 children were born with NTD in the East of Ireland (Table 5, Fig. 4). Overall, 51% (83/164) had spina bifida, 33% (54/164) had anencephaly and the remainder (16%) had encephalocoele. The overall rate was generally stable during the five-year period with some fluctuation in the SEHB.

Table 5. Neural Tube Defects: numbers and birth prevalence rates per 10,000 births 1997-2001

	EAST			ERHA			NEHB			SEHB		
YEAR	Births	No.	Rate /10,000	Births	No.	Rate /10,000	Births	No.	Rate /10,000	Births	No.	Rate /10,000
1997	**30,300**	**31**	**10.2**	20,285	20	9.9	4,532	6	13.2	5,483	5	9.1
1998	**31,092**	**29**	**9.3**	20,899	22	10.5	4,540	3	6.6	5,653	4	7.1
1999	**31,324**	**36**	**11.5**	20,767	25	12.0	4,786	6	12.5	5,771	5	8.7
2000	**31,861**	**34**	**10.7**	21,229	21	9.9	4,826	5	10.4	5,806	8	13.8
2001	**33,835**	**34**	**10.0**	22,137	20	9.0	5,378	6	11.2	6,320	8	12.7
Total	**158,412**	**164**	**10.4**	105,317	108	10.3	24,062	26	10.8	29,033	30	10.3

Fig. 4. Neural Tube defects: trends in birth prevalence rates per 10,000 births 1997-2001



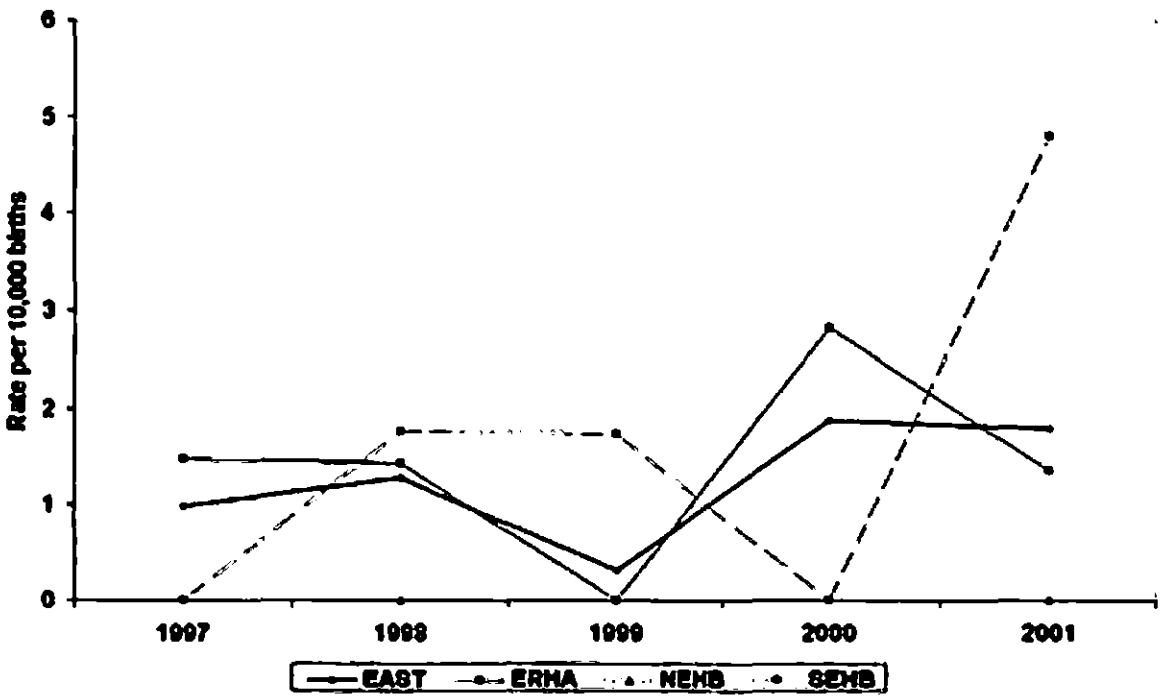
HOLOPROSENCEPHALY

Holoprosencephaly including arhinencephaly is relatively rare and occurs early in intrauterine life from failure of normal lobar development of the brain. The anomaly is frequently severe, resulting in intra-uterine death, although some may have near normal brain development with some facial deformities of the eye, nose and upper lip. The cause is largely unknown, however, there is a known association with specific chromosomal anomalies. A total of 20 cases of holoprosencephaly occurred in the East of Ireland from 1997-2001 (Table 6, Fig. 5). A slightly higher number in the year 2000 compared with earlier years was likely due to a higher number of children born with associated chromosomal anomalies.

Table 6. Holoprosencephaly: numbers and birth prevalence rates per 10,000 births 1997-2001

	EAST			ERHA			NEHB			SEHB		
YEAR	Births	No.	Rate /10,000	Births	No.	Rate /10,000	Births	No.	Rate /10,000	Births	No.	Rate /10,000
1997	30,300	3	1.0	20,285	3	1.5	4,532	0	0	5,483	0	0.0
1998	31,092	4	1.3	20,899	3	1.4	4,540	0	0	5,653	1	1.8
1999	31,324	1	0.3	20,767	0	0.0	4,786	0	0.0	5,771	1	1.7
2000	31,861	6	1.9	21,229	6	2.8	4,826	0	0.0	5,806	0	0.0
2001	33,835	6	1.8	22,137	3	1.4	5,378	0	0.0	6,320	3	4.8
Total	158,412	20	1.3	105,317	15	1.4	24,062	0	0.0	29,033	5	1.7

Fig. 5. Holoprosencephaly: trends in birth prevalence rates per 10,000 births 1997-2001



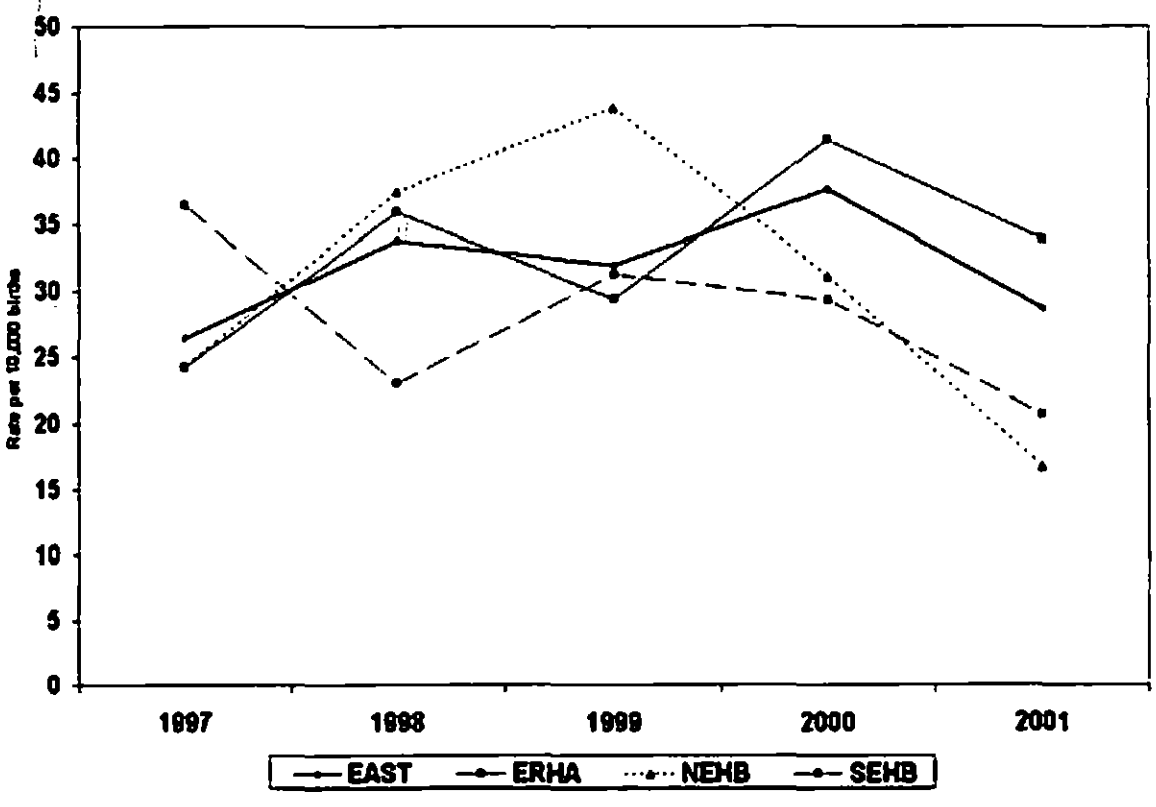
CHROMOSOMAL ANOMALIES

Chromosomal anomalies accounted for approximately 10% of all congenital anomalies during the study period. The most prevalent chromosomal anomalies are Down syndrome (Trisomy 21), Patau's syndrome (Trisomy 13) and Edward's syndrome (Trisomy 18), which together accounted for 90% of all chromosomal anomalies. The birth prevalence rate for all chromosomal anomalies combined fluctuated during the five-year period, with a peak in 2000 (Table 7, Fig. 6). The rate for all chromosomal anomalies follows the same pattern as that of Down syndrome, as 75% of all chromosomal anomalies were Down syndrome.

Table 7. All chromosomal anomalies: numbers and birth prevalence rates per 10,000 births 1997-2001

	EAST			ERHA			NEHB			SEHB		
YEAR	Births	No.	Rate /10,000	Births	No.	Rate /10,000	Births	No.	Rate /10,000	Births	No.	Rate /10,000
1997	30,300	80	26.4	20,285	49	24.2	4,532	11	24.3	5,483	20	36.5
1998	3,1092	105	33.8	20,899	75	35.9	4,540	17	37.4	5,653	13	23.0
1999	31,324	100	31.9	20,767	61	29.4	4,786	21	43.9	5,771	18	31.2
2000	31,861	120	37.7	21,229	88	41.5	4,826	15	31.1	5,806	17	13.8
2001	33,835	97	28.7	22,137	75	33.9	5,378	9	16.7	6,320	13	12.7
Total	158,412	502	31.7	105,317	348	33.0	24,062	73	30.3	29,033	81	27.9

Fig. 6. All chromosomal anomalies: trends in birth prevalence rates per 10,000 births 1997-2001



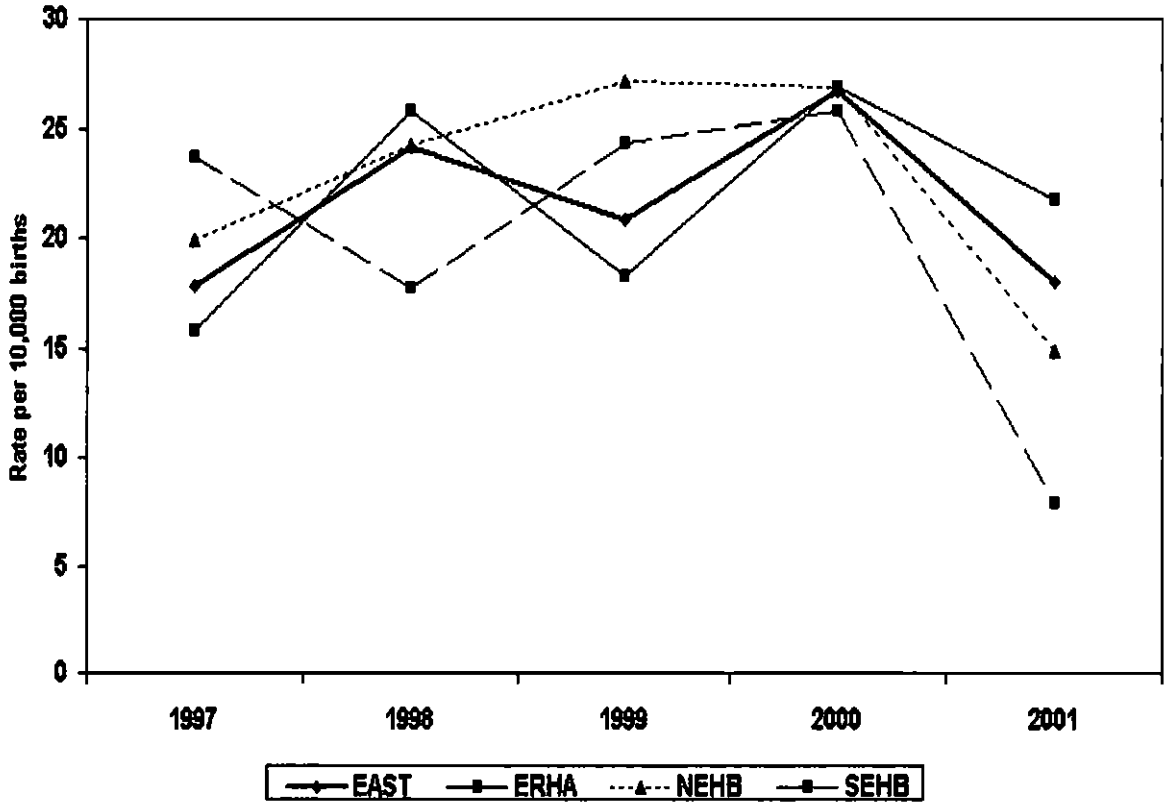
Trisomy 21 (Down syndrome)

More than 90% of Trisomy 21 is due to non-disjunction, resulting in three chromosome 21 in each cell rather than two. Approximately 5% is due to a translocation with chromosome 14 and the remainder are mosaic. Down syndrome is associated with a characteristic appearance and a range of other anomalies including mental retardation, heart defects and vision and hearing problems, with implications for a wide range of health and social services as well as the immediate family. Trisomy 21 is more likely to occur with increasing maternal age; the risk increases with age particularly over 30 years or more. However, children with Trisomy 21 can be born to mothers of any age. During the five-year period 1997-2001, there were 340 children born in the East of Ireland with Trisomy 21 (Table 8, Fig. 7), with the highest rates in 1998 and 2000 overall, but with variation within each health board.

Table 8. Trisomy 21: numbers and birth prevalence rates per 10,000 births 1997-2001

	EAST			ERHA			NEHB			SEHB		
YEAR	Births	No.	Rate /10,000	Births	No.	Rate 10,000	Births	No.	Rate /10,000	Births	No.	Rate /10,000
1997	**30,300**	**54**	**17.8**	20,285	32	15.8	4,532	9	19.9	5,483	13	23.7
1998	**31,092**	**75**	**24.1**	20,899	54	25.8	4,540	11	24.2	5,653	10	17.7
1999	**31,324**	**65**	**20.8**	20,767	38	18.3	4,786	13	27.2	5,771	14	24.3
2000	**31,861**	**85**	**26.7**	21,229	57	26.9	4,826	13	26.9	5,806	15	25.8
2001	**33,835**	**61**	**18.0**	22,137	48	21.7	5,378	8	14.9	6,320	5	7.9
Total	**158,412**	**340**	**21.5**	105,317	229	21.7	24,062	54	22.4	29,033	57	19.6

Fig. 7. Trisomy 21: trends in birth prevalence rates per 10,000 births 1997-2001



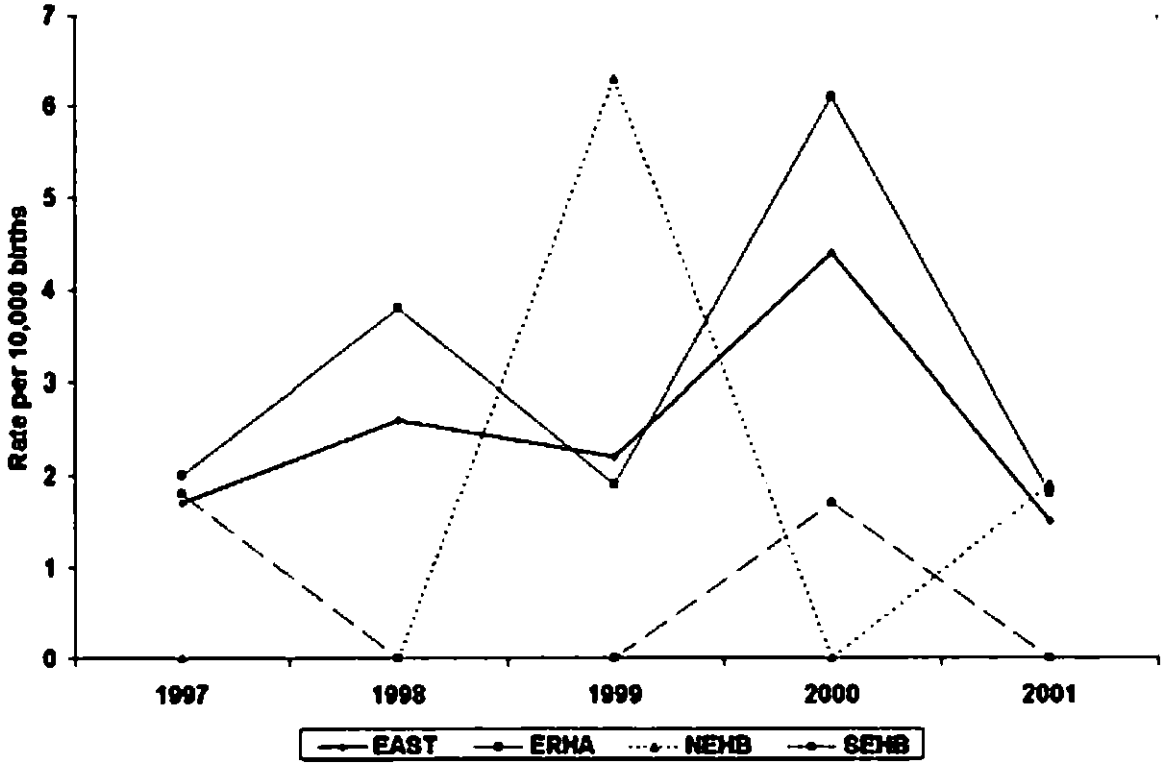
Trisomy 13 (Patau's syndrome)

Trisomy 13 is a severe chromosomal anomaly frequently incompatible with life. It is associated with a wide range of other anomalies, especially heart defects. As with Trisomy 21, the main known risk factor is advancing maternal age. Trisomy 13 is relatively rare and during the five-year period most of the 39 children were from the catchment of the ERHA (Table 9, Fig. 8). For reasons that are unclear, the rate was highest in 2000.

Table 9. Trisomy 13: numbers and birth prevalence rates per 10,000 births 1997-2001

	EAST			ERHA			NEHB			SEHB		
YEAR	Births	No.	Rate /10,000	Births	No.	Rate 10,000	Births	No.	Rate /10,000	Births	No.	Rate /10,000
1997	**30,300**	**5**	**1.7**	20,285	4	2.0	4,532	0	0.0	5,483	1	1.8
1998	**31,092**	**8**	**2.6**	20,899	8	3.8	4,540	0	0.0	5,653	0	0.0
1999	**31,324**	**7**	**2.2**	20,767	4	1.9	4,786	3	6.3	5,771	0	0.0
2000	**31,861**	**14**	**4.4**	21,229	13	6.1	4,826	0	0.0	5,806	1	1.7
2001	**33,835**	**5**	**1.5**	22,137	4	1.8	5,378	1	1.9	6,320	0	0.0
Total	**158,412**	**39**	**2.5**	105,317	33	3.1	24,062	4	1.7	29,033	2	0.7

Fig. 8. Trisomy 13: trends in birth prevalence rates per 10,000 births 1997-2001



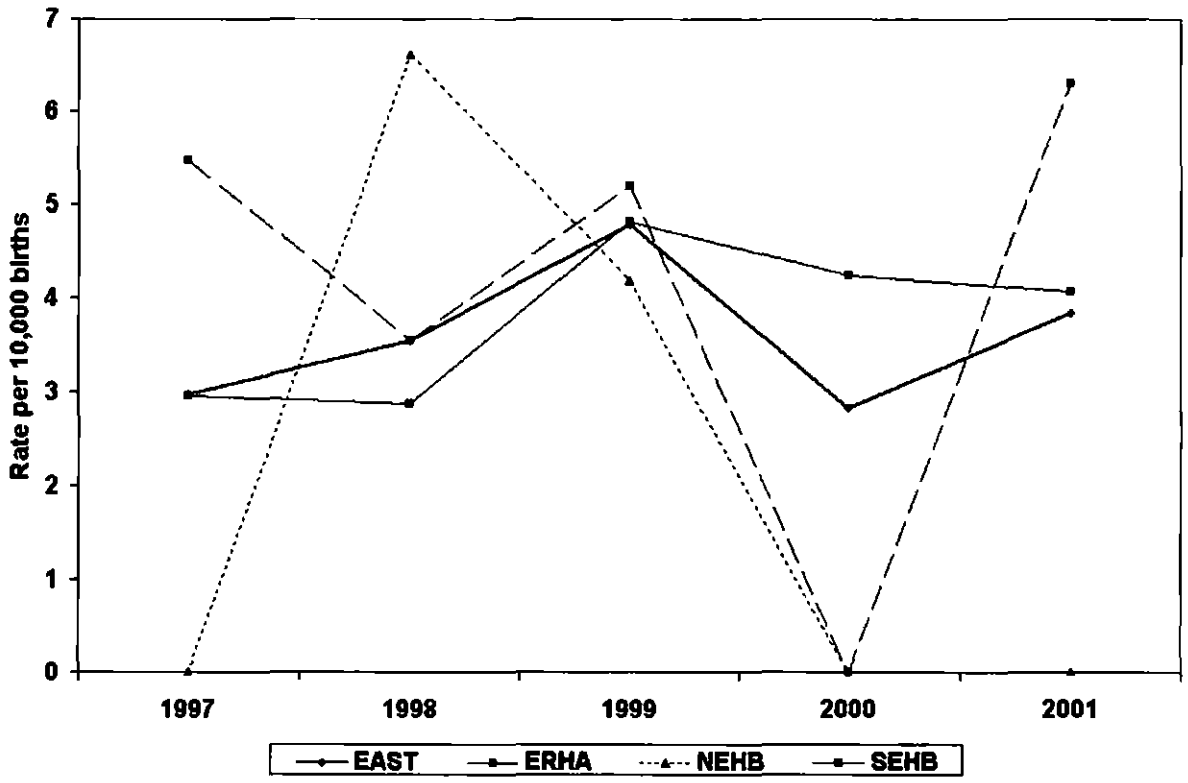
Trisomy 18 (Edward's syndrome)

Trisomy 18 is a severe chromosomal anomaly frequently incompatible with life. It is associated with a wide range of other anomalies, especially heart defects. As with Trisomy 21 and Trisomy 13, the main known risk factor is advancing maternal age. Trisomy 18 is slightly more prevalent than Trisomy 13. During the five-year period there were 57 children with Trisomy 18 (Table 10, Fig. 9), mostly in the ERHA catchment. Although the birth prevalence per 10,000 births varied between health boards/authority, the highest rates occurred in 1999 and 2001.

Table 10. Trisomy 18: numbers and birth prevalence rates per 10,000 births 1997-2001

	EAST			ERHA			NEHB			SEHB		
YEAR	Births	No.	Rate /10,000	Births	No.	Rate /10,000	Births	No.	Rate /10,000	Births	No.	Rate /10,000
1997	**30,300**	**9**	**3.0**	20,285	6	3.0	4,532	0	0.0	5,483	3	5.5
1998	**31,092**	**11**	**3.5**	20,899	6	2.9	4,540	3	6.6	5,653	2	3.5
1999	**31,324**	**15**	**4.8**	20,767	10	4.8	4,786	2	4.2	5,771	3	5.2
2000	**31,861**	**9**	**2.8**	21,229	9	4.2	4,826	0	0.0	5,806	0	0.0
2001	**33,835**	**13**	**3.8**	22,137	9	4.1	5,378	0	0.0	6,320	4	6.3
Total	**158,412**	**57**	**3.6**	105,317	40	3.8	24,062	5	2.1	29,033	12	4.1

Fig. 9. Trisomy 18: trends in birth prevalence rates per 10,000 births 1997-2001



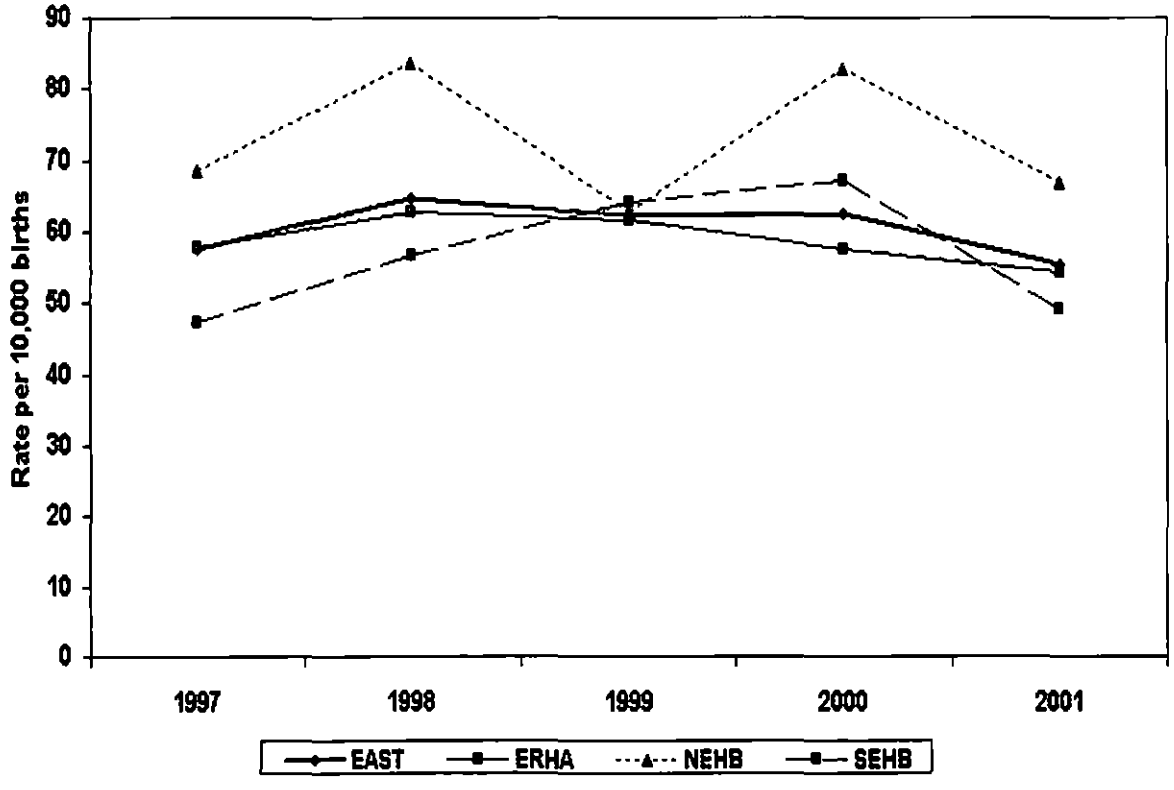
CONGENITAL HEART DISEASE

Congenital heart anomalies accounted for 20% of all congenital anomalies during the study period. The cause of congenital heart defects is unknown, although some may be associated with factors such as rubella or diabetes. A high proportion of children born with chromosomal anomalies also have a congenital heart defect. Specific anomalies analysed were hypoplastic left heart, coarctation of the aorta, transposition of the great vessels and Fallot's tetralogy. There were 958 children with congenital heart anomalies born in the east of the country during the five-year period (Table 11, Fig. 10). The birth prevalence rate for all congenital heart anomalies remained stable at approximately 60 per 10,000 births. Rates were slightly higher in the north-eastern counties.

Table 11. All heart anomalies: numbers and birth prevalence rates per 10,000 births 1997-2001

	EAST			ERHA			NEHB			SEHB		
YEAR	Births	No.	Rate /10,000	Births	No.	Rate /10,000	Births	No.	Rate /10,000	Births	No.	Rate /10,000
1997	30,300	174	57.4	20,285	117	57.7	4,532	31	68.4	5,483	26	47.4
1998	31,092	201	64.6	20,899	131	62.7	4,540	38	83.7	5,653	32	56.6
1999	31,324	195	62.3	20,767	128	61.6	4,786	30	62.7	5,771	37	64.1
2000	31,861	201	63.1	21,229	122	57.5	4,826	40	82.9	5,806	39	67.2
2001	33,835	187	55.3	22,137	120	54.2	5,378	36	66.9	6,320	31	49.1
Total	158,412	958	60.5	105,317	618	58.7	24,062	175	72.7	29,033	165	56.8

Fig. 10. Congenital heart defects: trends in birth prevalence rates per 10,000 births 1997-2001



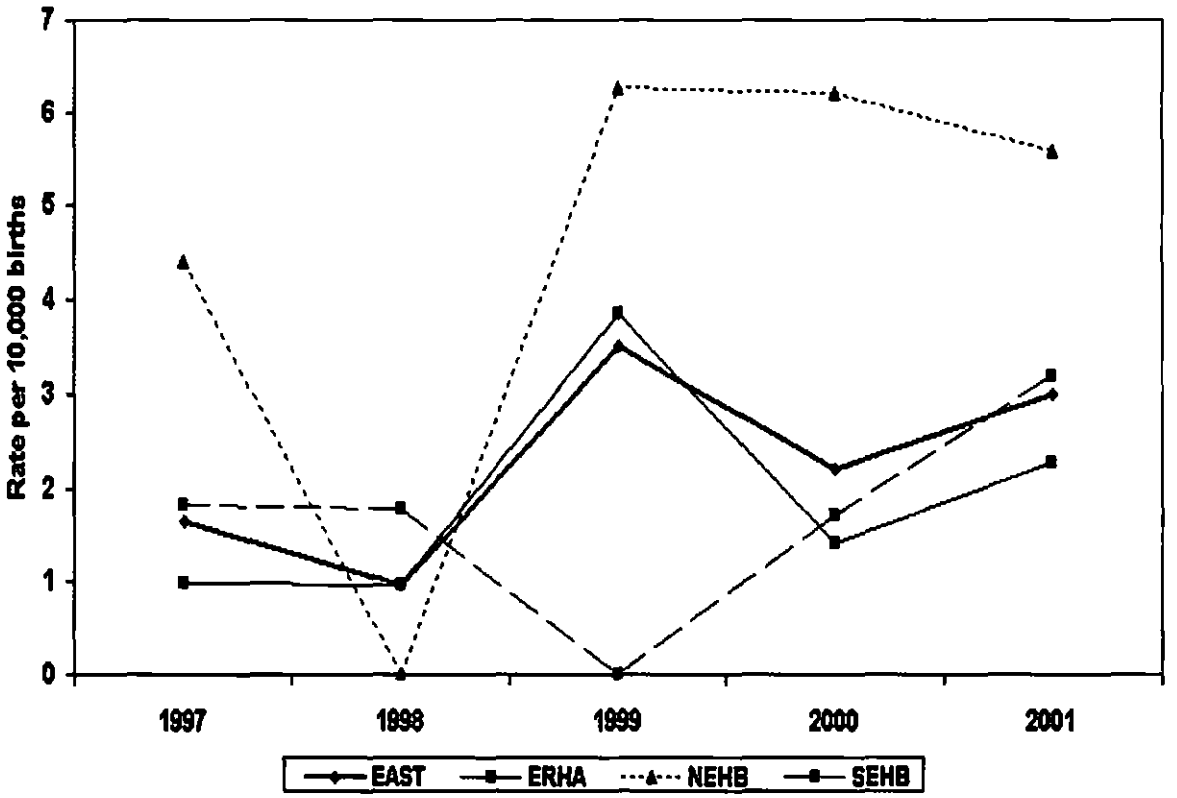
Hypoplastic left heart

This relatively rare anomaly is a result of failure of development of the left side of the heart in intrauterine life. It is usually a lethal condition, as it is extremely difficult to treat. There were 36 children born with hypoplastic left heart during the five-year period (Table 12, Fig. 11). The birth prevalence rate fluctuated, the average being 2.2 per 10,000 births per year. The highest overall rate occurred in 1999. Rates were higher in the north-eastern counties from 1999.

Table 12. Hypoplastic Left Heart: numbers and birth prevalence rates per 10,000 births1997-2001

	EAST			ERHA			NEHB			SEHB		
YEAR	Births	No.	Rate /10,000	Births	No.	Rate /10,000	Births	No.	Rate /10,000	Births	No.	Rate /10,000
1997	30,300	5	1.7	20,285	2	1.0	4,532	2	4.4	5,483	1	1.8
1998	31,092	3	1.0	20,899	2	1.0	4,540	0	0.0	5,653	1	1.8
1999	31,324	11	3.5	20,767	8	3.9	4,786	3	6.3	5,771	0	0.0
2000	31,861	7	2.2	21,229	3	1.4	4,826	3	6.2	5,806	1	1.7
2001	33,835	10	2.7	22,137	5	2.3	5,378	3	5.6	6,320	2	3.2
Total	158,412	36	2.2	105,317	20	1.9	24,062	11	4.6	29,033	5	1.7

Fig. 11. Hypoplastic Left Heart: trends in birth prevalence rates per 10,000 births 1997-2001



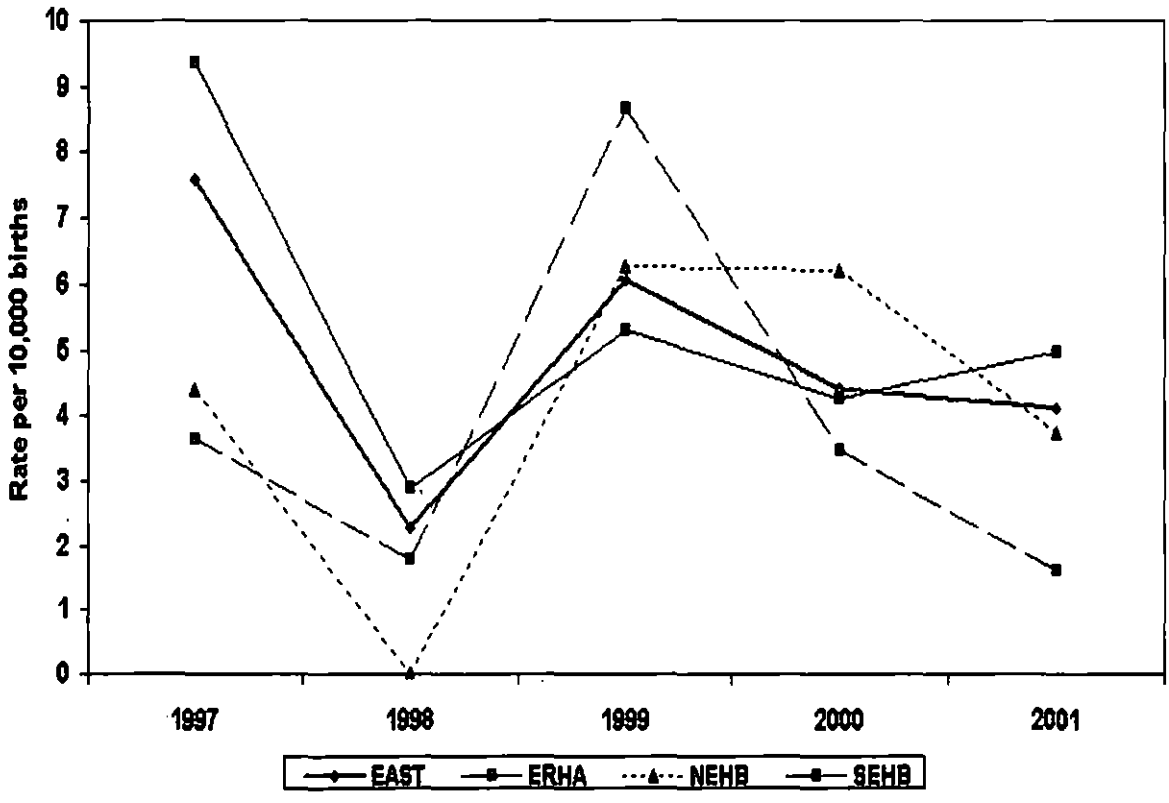
Transposition of the Great Vessels

Transposition of the Great Vessels is a common cause of congenital cyanotic heart disease, in which the two major vessels that carry blood away from the heart, the aorta and the pulmonary artery, are transposed. This can occur with or without other cardiac defects; the severity can therefore vary. Although the cause is largely unknown, there may be an association with some maternal factors such as rubella or diabetes, as in other heart defects. Early treatment is necessary, the outcome depending on the severity of the anomaly. There were 77 children born with this anomaly during the five-year period. The average birth prevalence rate per 10,000 births was 4.8 (Table 13, Fig. 12), although there was fluctuation from year to year, with the highest rate in 1997.

Table 13: Transposition Great Vessels: numbers and birth prevalence rates per 10,000 births 1997-2001

	EAST			ERHA			NEHB			SEHB		
YEAR	Births	No.	Rate /10,000	Births	No.	Rate /10,000	Births	No.	Rate /10,000	Births	No.	Rate /10,000
1997	**30,300**	**23**	**7.6**	20,285	19	9.4	4,532	2	4.4	5,483	2	3.6
1998	**31,092**	**7**	**2.3**	20,899	6	2.9	4,540	0	0.0	5,653	1	1.8
1999	**31,324**	**19**	**6.1**	20,767	11	5.3	4,786	3	6.3	5,771	5	8.7
2000	**31,861**	**14**	**4.4**	21,229	9	4.2	4,826	3	6.2	5,806	2	3.4
2001	**33,835**	**14**	**4.1**	22,137	11	5.0	5,378	2	3.7	6,320	1	1.6
Total	**158,412**	**77**	**4.9**	105,317	56	5.3	24,062	10	4.2	29,033	11	3.8

Fig. 12. Transposition Great Vessels: trends in birth prevalence rates per 10,000 births 1997-2001



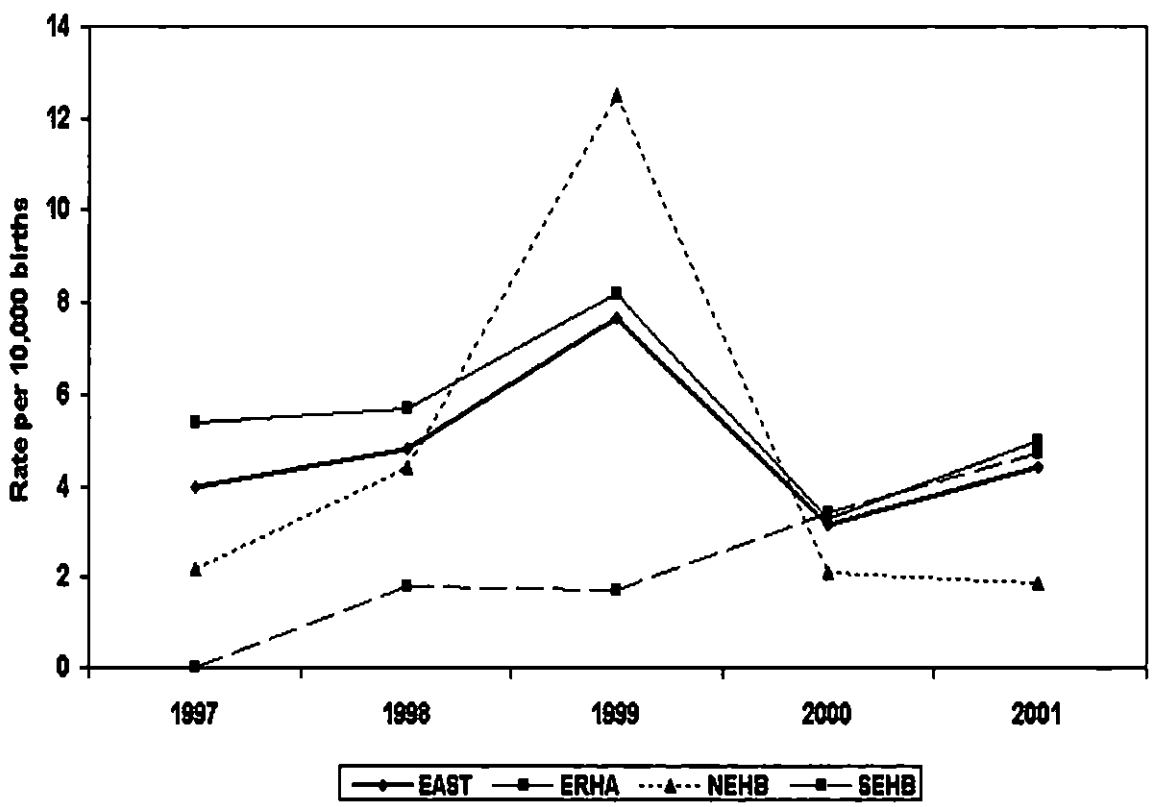
Coarctation of the Aorta

In coarctation of the aorta, there is narrowing of the aorta somewhere along its length, usually at the insertion of the ductus arteriosus. It is more common with some genetic conditions e.g. Turner's syndrome. It may be diagnosed soon after birth or later in childhood and surgery is usually necessary. There were 76 children born with coarctation of the aorta during the five-year period. The average birth prevalence rate per 10,000 births was 4.7 (Table 14, Fig.13), although there was fluctuation from year to year. The highest overall rate occurred in 1999, with the highest number in that year in the ERHA catchment and the NEHB.

Table 14. Coarctation of the Aorta: numbers and birth prevalence rates per10,000 births 1997-2001

	EAST			ERHA			NEHB			SEHB		
YEAR	Births	No.	Rate /10,000	Births	No.	Rate /10,000	Births	No.	Rate /10,000	Births	No.	Rate /10,000
1997	30,300	12	4.0	20,285	11	5.4	4,532	1	2.2	5,483	0	0.0
1998	31,092	15	4.8	20,899	12	5.7	4,540	2	4.4	5,653	1	1.8
1999	31,324	24	7.7	20,767	17	8.2	4,786	6	12.5	5,771	1	1.7
2000	31,861	10	3.1	21,229	7	3.3	4,826	1	2.1	5,806	2	3.4
2001	33,835	15	4.4	22,137	11	5.0	5,378	1	1.9	6,320	3	4.7
Total	158,412	76	4.8	105,317	58	5.5	24,062	11	4.6	29,033	7	2.4

Fig. 13. Coarctation of the Aorta: trends in birth prevalence rates per10,000 births 1997-2001



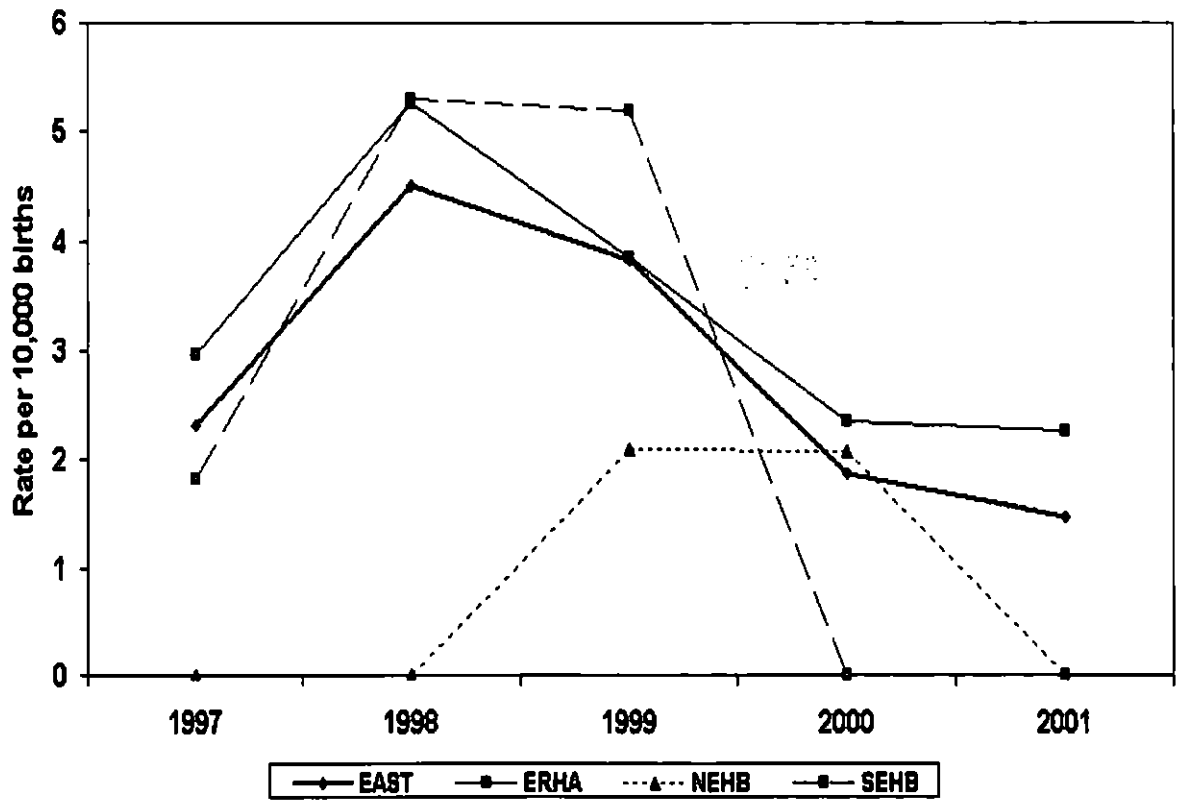
Tetralogy of Fallot

Tetralogy of Fallot is a cause of congenital cyanotic heart disease. Although the aetiology is unknown, there may be an association with some maternal factors such as rubella or diabetes, as in other heart defects. It is also more common in children with Trisomy 21. It is comprised of four anomalies in the heart: defects of the ventricular septum, the aorta, pulmonary artery and right ventricle. Surgical repair in early life is necessary and the prognosis is good. There were 44 children born with this anomaly during the five-year period. The overall average birth prevalence rate per 10,000 births was 2.8 (Table 15, Fig.14), although with fluctuation from year to year. The highest overall rate occurred in 1998, and corresponds with a higher birth prevalence of Trisomy 21 during that year.

Table 15. Tetralogy of Fallot: numbers and birth prevalence rates per 10,000 births 1997-2001

	EAST			ERHA			NEHB			SEHB		
YEAR	Births	No.	Rate /10,000	Births	No.	Rate /10,000	Births	No.	Rate /10,000	Births	No.	Rate /10,000
1997	**30,300**	**7**	**2.3**	20,285	6	3.0	4,532	0	0.0	5,483	1	1.8
1998	**31,092**	**14**	**4.5**	20,899	11	5.3	4,540	0	0.0	5,653	3	5.3
1999	**31,324**	**12**	**3.8**	20,767	8	3.9	4,786	1	2.1	5,771	3	5.2
2000	**31,861**	**6**	**1.9**	21,229	5	2.4	4,826	1	2.1	5,806	0	0.0
2001	**33,835**	**5**	**1.5**	22,137	5	2.3	5,378	0	0.0	6,320	0	0.0
Total	**158,412**	**44**	**2.8**	105,317	35	3.3	24,062	2	0.8	29,033	7	2.4

Fig. 14. Tetralogy of Fallot: trends in birth prevalence rates per 10,000 births 1997-2001



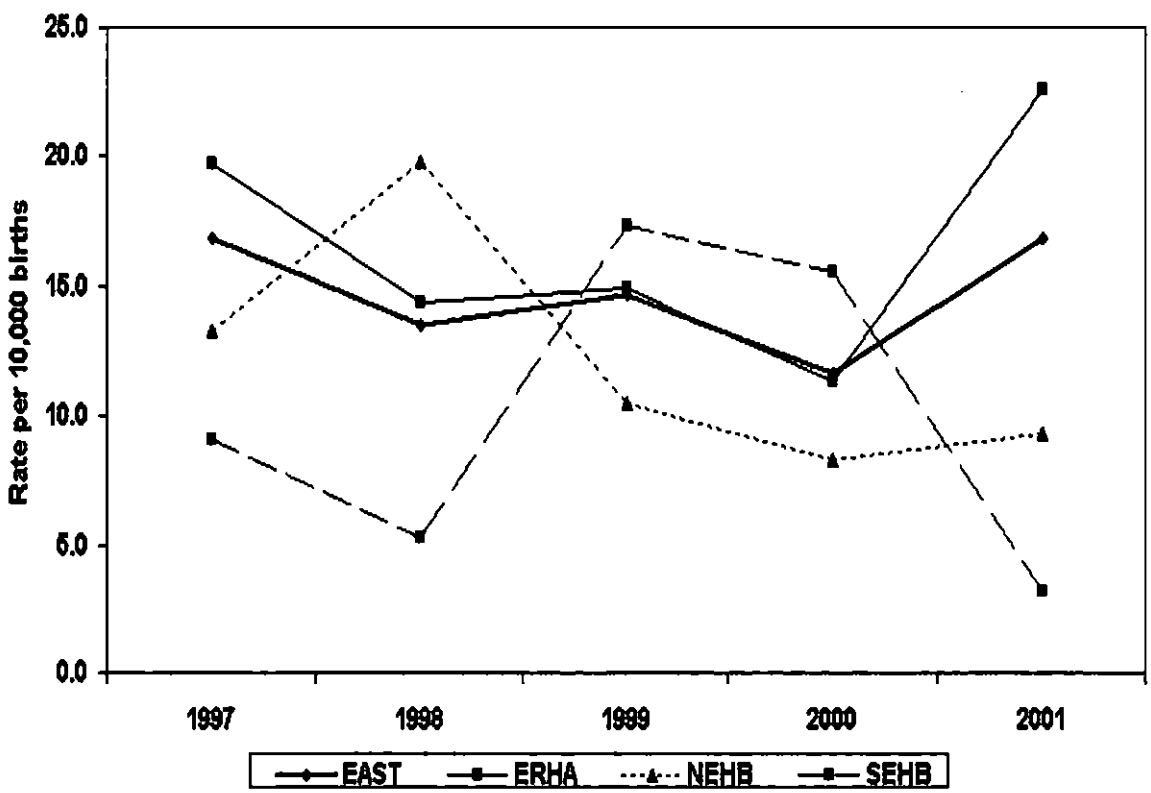
CLEFT ANOMALIES

This group of anomalies is comprised of isolated cleft lip, isolated cleft palate and cleft lip with cleft palate. The anomalies may range from a small defect in the lip to a complete fissure extending into the roof of the mouth and nose; these features may occur separately or together. A number of factors have been associated with cleft anomalies, including genetic and environmental factors (e.g. teratogens, infections). Depending on the extent of the defect, surgical treatment is frequently required in early life. There were 232 children born with cleft anomalies in the East of Ireland during the five-year period. The average birth prevalence rate per 10,000 births for all types of cleft anomaly was 14.7 (Table 16, Fig. 15), with fluctuation from year to year. The highest overall rates occurred in 1997 and 2001, although the years with the highest rates tended to be in different years among the health boards / authority.

Table 16. Cleft Lip and/or palate: numbers and birth prevalence rates per 10,000 births 1997-2001

	EAST			ERHA			NEHB			SEHB		
YEAR	Births	No.	Rate /10,000	Births	No.	Rate /10,000	Births	No.	Rate /10,000	Births	No.	Rate /10,000
1997	**30,300**	**51**	**16.8**	20,285	40	19.7	4,532	6	13.2	5,483	5	9.1
1998	**31,092**	**42**	**13.5**	20,899	30	14.4	4,540	9	19.8	5,653	3	5.3
1999	**31,324**	**46**	**14.7**	20,767	31	14.9	4,786	5	10.4	5,771	10	17.3
2000	**31,861**	**37**	**11.6**	21,229	25	11.3	4,826	4	8.3	5,806	9	15.5
2001	**33,835**	**56**	**16.6**	22,137	49	22.1	5,378	5	9.3	6,320	2	3.2
Total	**158,412**	**232**	**14.7**	105,317	174	16.5	24,062	29	12.1	29,033	29	10.0

Fig. 15. Cleft Lip &/or palate: trends in birth prevalence rates per 10,000 births 1997-2001



DIGESTIVE SYSTEM ANOMALIES

The main digestive system anomalies studied were tracheo-oesophageal fistula, atresia of small intestine and ano-rectal atresia.

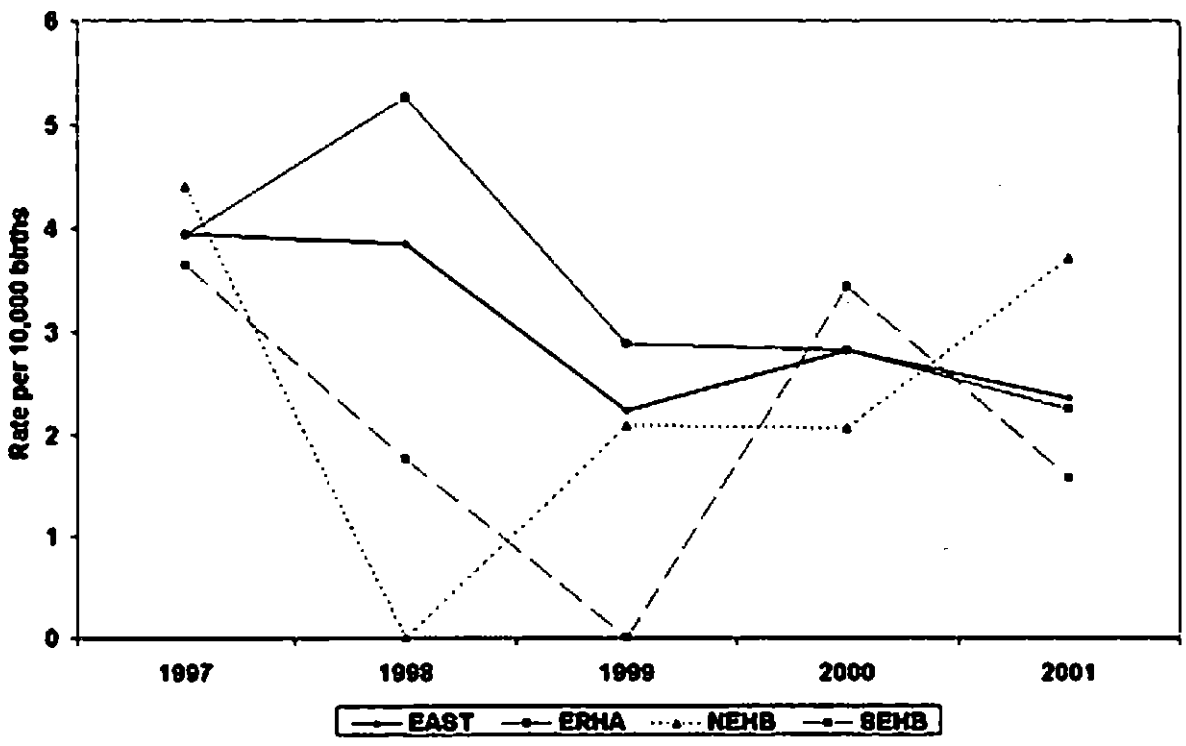
Tracheo-oesophageal fistula

This is characterised by a communication between the trachea and the oesophagus, and usually occurs in association with oesophageal atresia. There are frequently other abnormalities, e.g. cardiac, genitourinary, chromosomal abnormalities (trisomies 18, 21, 13). Tracheo-oesophageal fistula may result in life-threatening complications neonatally and surgical repair is usually undertaken within a few days of birth. Prognosis is very good, although it depends on the presence and type of other abnormalities. There were 48 children born with tracheo-oesophageal fistula during the five-year period. Overall, the average birth prevalence rate per 10,000 births was 3.0 (Table 17, Fig. 16). The highest rates occurred in 1997 and 1998, although varying from year to year among the health boards/authority.

Table 17 Tracheo-oesophageal fistula: numbers and birth prevalence rates per 10,000 births 1997-2001

	EAST			ERHA			NEHB			SEHB		
YEAR	Births	No.	Rate /10,000	Births	No.	Rate /10,000	Births	No.	Rate /10,000	Births	No.	Rate /10,000
1997	**30,300**	**12**	**4.0**	20,285	8	3.9	4,532	2	4.4	5,483	2	3.6
1998	**31,092**	**12**	**3.9**	20,899	11	5.3	4,540	0	0.0	5,653	1	1.8
1999	**31,324**	**7**	**2.2**	20,767	6	2.9	4,786	1	2.1	5,771	0	0.0
2000	**31,861**	**9**	**2.8**	21,229	6	2.8	4,826	1	2.1	5,806	2	3.4
2001	**33,835**	**8**	**2.4**	22,137	5	2.3	5,378	2	3.7	6,320	1	1.6
Total	**158,412**	**48**	**3.0**	105,317	36	3.4	24,062	6	2.5	29,033	6	2.1

Fig. 16. Tracheo-oesophageal fistula: trends in birth prevalence rates per 10,000 births 1997-2001



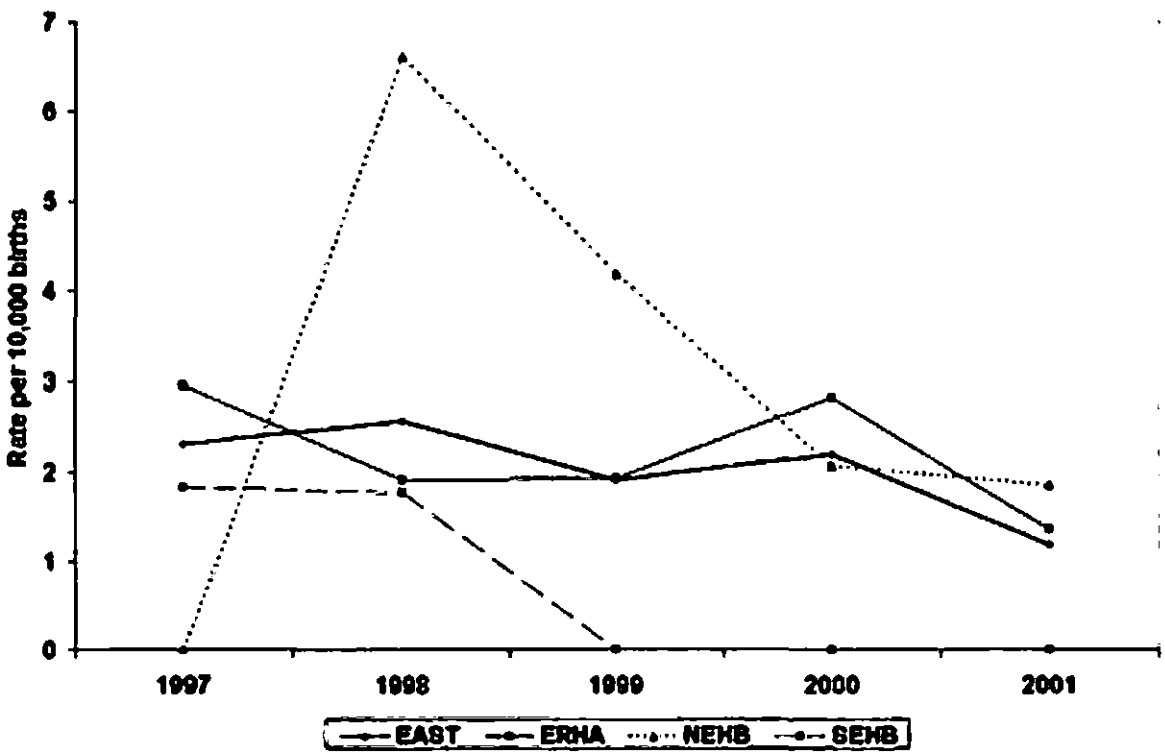
Intestinal atresia or stenosis

In intestinal atresia/stenosis there is complete or partial occlusion of the lumen of a segment of the small intestine. There are frequently associated abnormalities e.g. cardiac, genitourinary, chromosomal abnormalities (trisomies 18, 21, 13). Surgical treatment is usually required soon after birth. The prognosis depends on the anomaly itself and the presence of associated anomalies. There were 32 children born with intestinal atresia/stenosis during the five-year period. The average birth prevalence rate per 10,000 births was 2.0 (Table 18, Fig. 17). Overall, the rate was stable throughout the period, although higher rates were observed in the NEHB in 1998 and 1999.

Table 18 Intestinal atresia / stenosis: numbers and birth prevalence rates per 10,000 births 1997-2001

	EAST			ERHA			NEHB			SEHB		
YEAR	Births	No.	Rate /10,000	Births	No.	Rate /10,000	Births	No.	Rate /10,000	Births	No.	Rate /10,000
1997	**30,300**	**7**	**2.3**	20,285	6	3.0	4,532	0	0.0	5,483	1	1.8
1998	**31,092**	**8**	**2.6**	20,899	4	1.9	4,540	3	6.6	5,653	1	1.8
1999	**31,324**	**6**	**1.9**	20,767	4	1.9	4,786	2	4.2	5,771	0	0.0
2000	**31,861**	**7**	**2.2**	21,229	6	2.8	4,826	1	2.1	5,806	0	0.0
2001	**33,835**	**4**	**1.2**	22,137	3	1.4	5,378	1	1.9	6,320	0	0.0
Total	**158,412**	**32**	**2.0**	105,317	23	2.2	24,062	7	2.9	29,033	2	0.7

Fig 17. Intestinal atresia / stenosis: trends in birth prevalence rates per 10,000 births 1997-2001



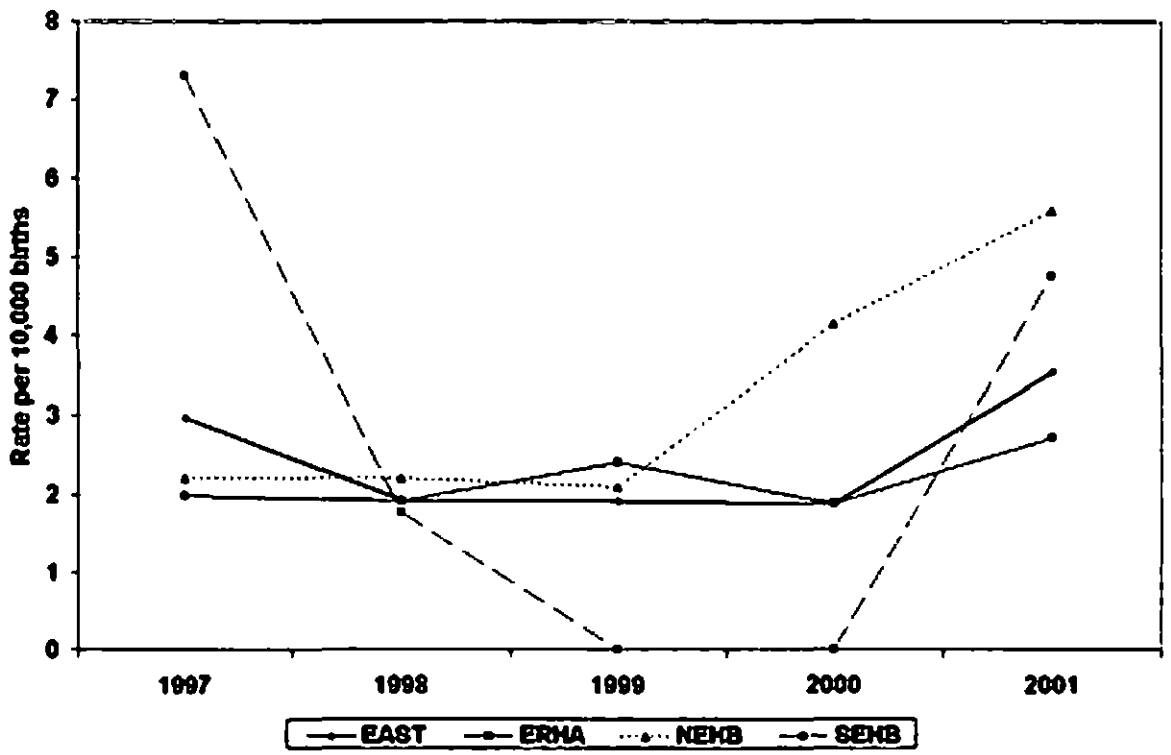
Ano-rectal atresia

Ano-rectal atresia is characterised by absence of continuity of the ano-rectal canal, or a communication between rectum and anus, or narrowing of the anal canal. Often there is a fistula with the genitourinary organs and sometimes a range of other anomalies. Surgical treatment is required soon after birth and the outcome is usually very good, depending on co-morbidities. There were 39 children born with ano-rectal atresia during the five-year period. The overall average birth prevalence rate per 10,000 births was 2.5 (Table 19, Fig. 18), with some fluctuation from year to year. The years with the highest rates varied among the health boards / authority.

Table 19. Ano-rectal atresia: numbers and birth prevalence rates per 10,000 births 1997-2001

	EAST			ERHA			NEHB			SEHB		
YEAR	Births	No.	Rate /10,000	Births	No.	Rate /10,000	Births	No.	Rate /10,000	Births	No.	Rate /10,000
1997	**30,300**	**9**	**3.0**	20,285	4	2.0	4,532	1	2.2	5,483	4	7.3
1998	**31,092**	**6**	**1.9**	20,899	4	1.9	4,540	1	2.2	5,653	1	1.8
1999	**31,324**	**6**	**1.9**	20,767	5	2.4	4,786	1	2.1	5,771	0	0.0
2000	**31,861**	**6**	**1.9**	21,229	4	1.9	4,826	2	4.1	5,806	0	0.0
2001	**33,835**	**12**	**3.5**	22,137	6	2.7	5,378	3	5.6	6,320	3	4.7
Total	**158,412**	**39**	**2.5**	105,317	23	2.2	24,062	8	3.3	29,033	8	2.8

Fig 18. Ano-rectal atresia: trends in birth prevalence rates per 10,000 births 1997-2001



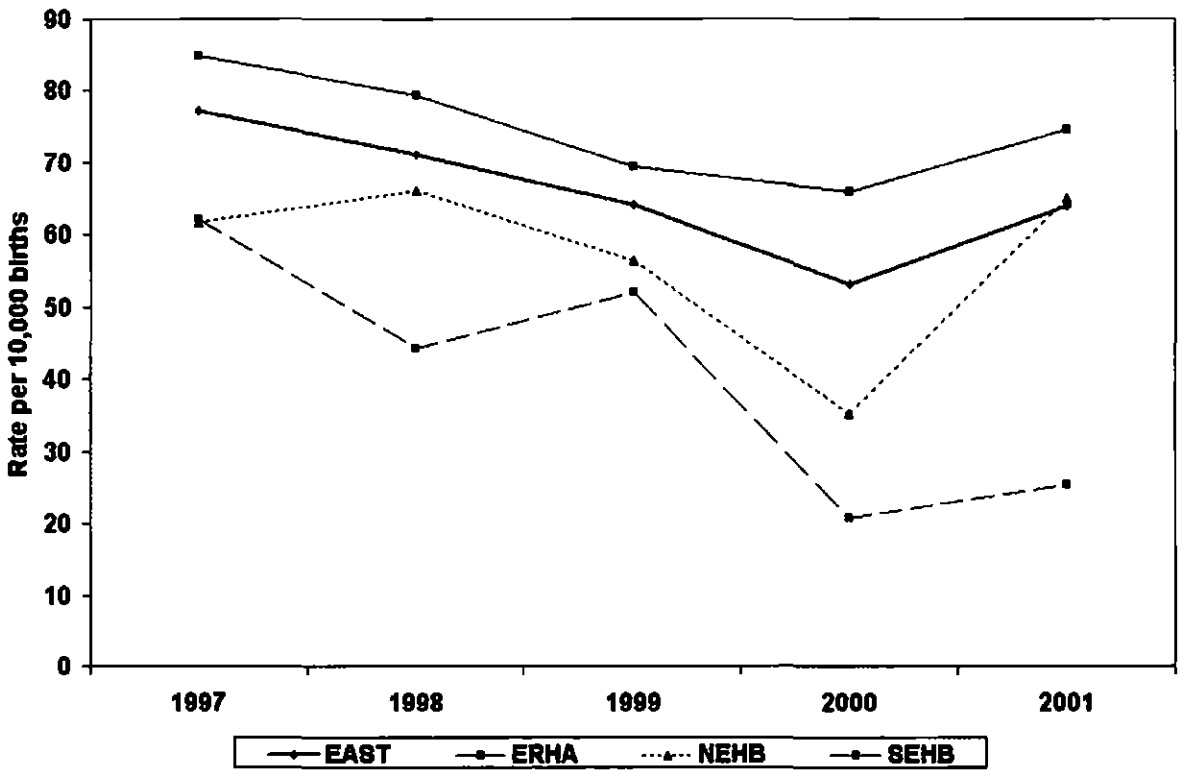
LIMB ANOMALIES

Limb anomalies are very common. They may consist of an absence of a limb or limb part, or extra or malformed parts. Consequently, there is a wide range of limb anomalies that can occur, and one or more limbs may be affected. The cause is multi-factorial, and treatment depends on the type of anomaly and whether other associated anomalies are present. There were 1,041 children born with limb anomalies during the five-year period. The average birth prevalence rate per 10,000 births was 65.7 (Table 20, Fig. 19). The rate fluctuated from year to year, with higher rates in the catchment of the ERHA. The lower overall rates in the south-eastern counties are likely to a result of incomplete ascertainment in 2000 and 2001.

Table 20. All Limb anomalies: numbers and birth prevalence rates per 10,000 births 1997-2001

	EAST			ERHA			NEHB			SEHB		
YEAR	Births	No.	Rate /10,000	Births	No.	Rate /10,000	Births	No.	Rate /10,000	Births	No.	Rate /10,000
1997	**30,300**	**234**	**77.2**	20,285	172	84.8	4,532	28	61.8	5,483	34	62.0
1998	**31,092**	**221**	**71.1**	20,899	166	79.4	4,540	30	66.1	5,653	25	44.2
1999	**31,324**	**201**	**64.2**	20,767	144	69.3	4,786	27	56.4	5,771	30	52.0
2000	**31,861**	**169**	**53.0**	21,229	140	65.9	4,826	17	35.2	5,806	12	20.7
2001	**33,835**	**216**	**63.8**	22,137	165	74.5	5,378	35	65.1	6,320	16	25.3
Total	**158,412**	**1,041**	**65.7**	105,317	787	74.7	24,062	137	56.9	29,033	117	40.3

Fig. 19. All Limb anomalies: trends in birth prevalence rates per 10,000 births 1997-2001



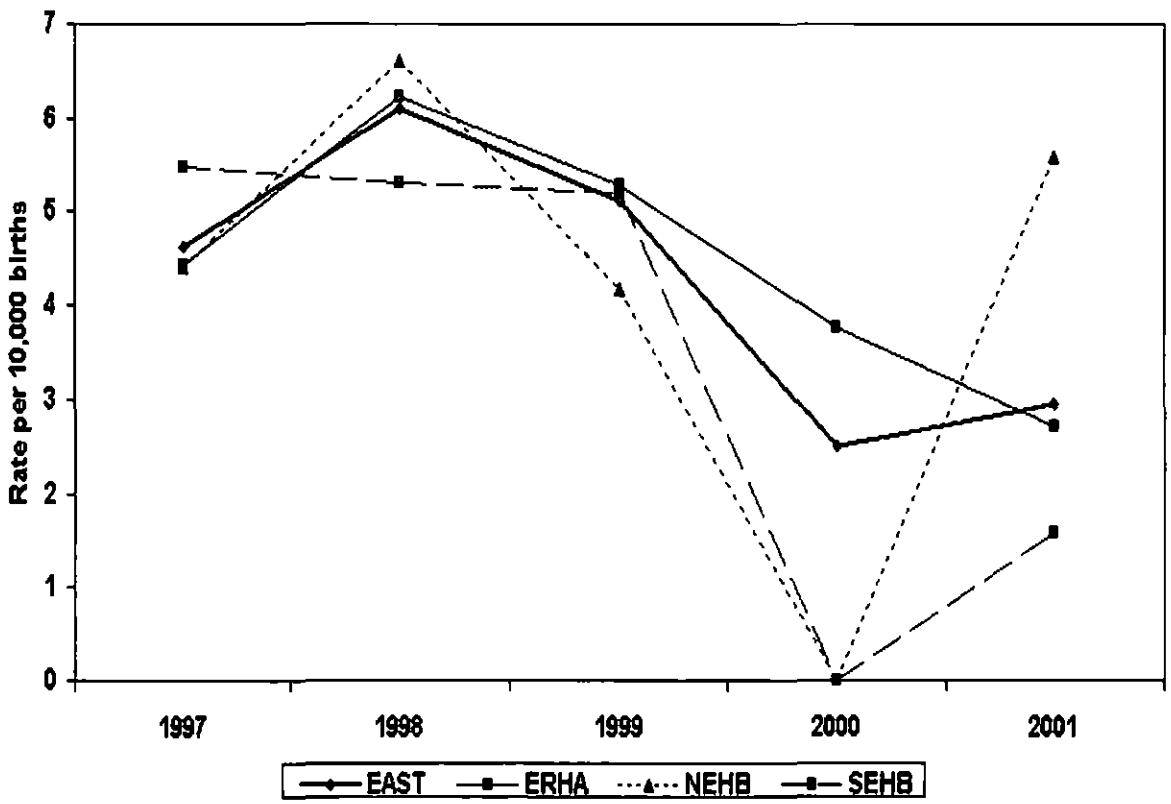
Limb reduction anomalies

Limb reduction anomalies are an important sub-group of limb anomalies and are characterised by total or partial absence, or severe underdevelopment of skeletal structures of the limbs. Approximately two-thirds are anomalies of the upper limbs. Limb reduction anomalies may occur simultaneously in the upper and lower limbs. There were 67 children born with limb anomalies during the five-year period. The average birth prevalence rate per 10,000 births was 4.2 (Table 21, Fig. 20). The rate fluctuated from year to year, with higher rates in the ERHA catchment. The highest overall rate was in 1998.

Table 21 Limb reduction anomalies: numbers and birth prevalence rates per 10,000 births 1997-2001

	EAST			ERHA			NEHB			SEHB		
YEAR	Births	No.	Rate /10,000	Births	No.	Rate /10,000	Births	No.	Rate /10,000	Births	No.	Rate /10,000
1997	**30,300**	**14**	**4.6**	20,285	9	4.4	4,532	2	4.4	5,483	3	5.5
1998	**31,092**	**19**	**6.1**	20,899	13	6.2	4,540	3	6.6	5,653	3	5.3
1999	**31,324**	**16**	**5.1**	20,767	11	5.3	4,786	2	4.2	5,771	3	5.2
2000	**31,861**	**8**	**2.5**	21,229	8	3.8	4,826	0	0.0	5,806	0	0.0
2001	**33,835**	**10**	**3.0**	22,137	6	2.7	5,378	3	5.6	6,320	1	1.6
Total	**158,412**	**67**	**4.2**	105,317	47	4.5	24,062	10	4.2	29,033	10	3.4

Fig. 20. Limb reduction anomalies: trends in birth prevalence rates per 10,000 births 1997-2001



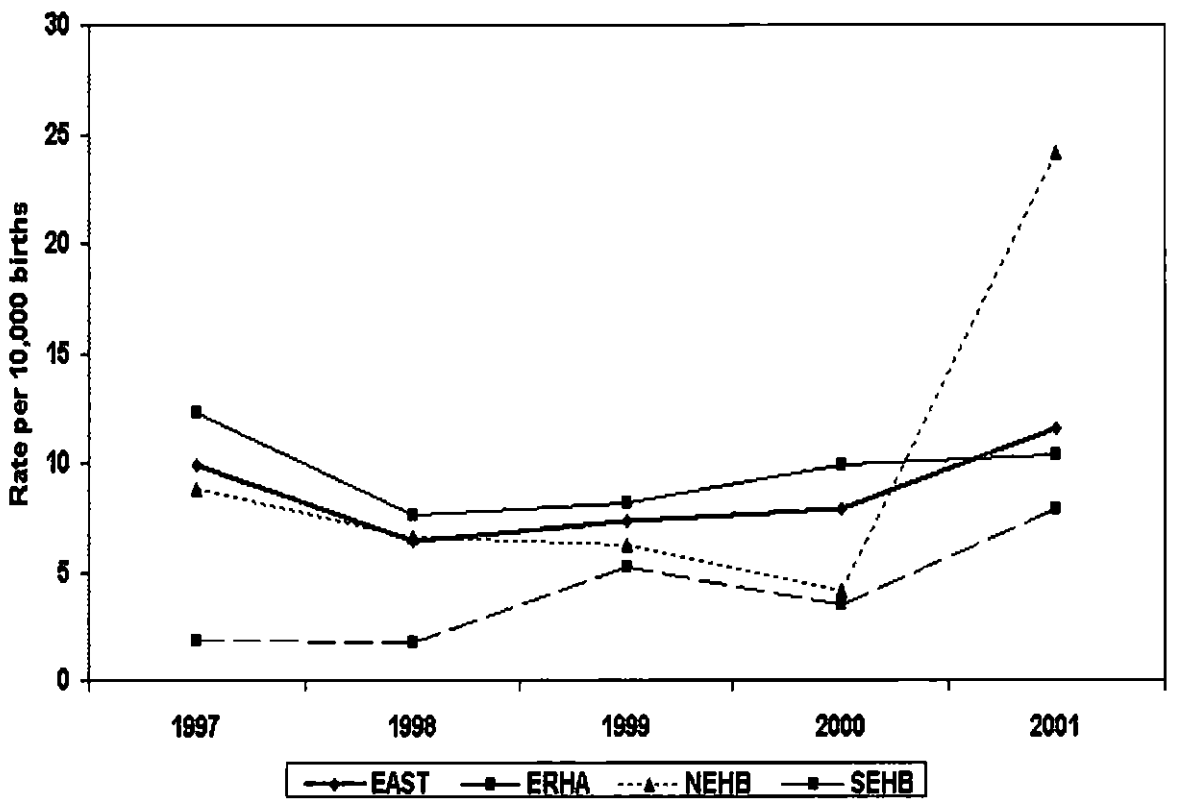
Polydactyly

Polydactyly is a common limb anomaly and consists of having more than five digits and can affect the hand, the foot, or both. Extra digits may be extremely rudimentary and attached by a small stalk (generally on the little finger side of the hand) or fairly well-formed and even functional. Rudimentary digits are generally removed. Sometimes polydactyly is familial or can be associated with a number of genetic syndromes. There were 139 children born with polydactyly during the five-year period. The average birth prevalence rate per 10,000 births was 8.8 (Table 22, Fig. 21); the rate was generally stable from year to year between 1997-2000. However, it rose in 2001, particularly in the NEHB and less so the SEHB.

Table 22. Polydactyly: numbers and birth prevalence rates per 10,000 births 1997-2001

	EAST			ERHA			NEHB			SEHB		
YEAR	Births	No.	Rate /10,000	Births	No.	Rate /10,000	Births	No.	Rate /10,000	Births	No.	Rate /10,000
1997	**30,300**	**30**	**9.9**	20,285	25	12.3	4,532	4	8.8	5,483	1	1.8
1998	**31,092**	**20**	**6.4**	20,899	16	7.7	4,540	3	6.6	5,653	1	1.8
1999	**31,324**	**23**	**7.3**	20,767	17	8.2	4,786	3	6.3	5,771	3	5.2
2000	**31,861**	**25**	**7.8**	21,229	21	9.9	4,826	2	4.1	5,806	2	3.4
2001	**33,835**	**41**	**12.1**	22,137	23	10.4	5,378	13	24.2	6,320	5	7.9
Total	**158,412**	**139**	**8.8**	105,317	102	9.7	24,062	25	10.4	29,033	12	4.1

Fig. 21. Polydactyly: trends in birth prevalence rates per 10,000 births 1997-2001



ABDOMINAL WALL DEFECTS

The two main abdominal wall defects are gastroschisis and omphalocoele (exomphalos).

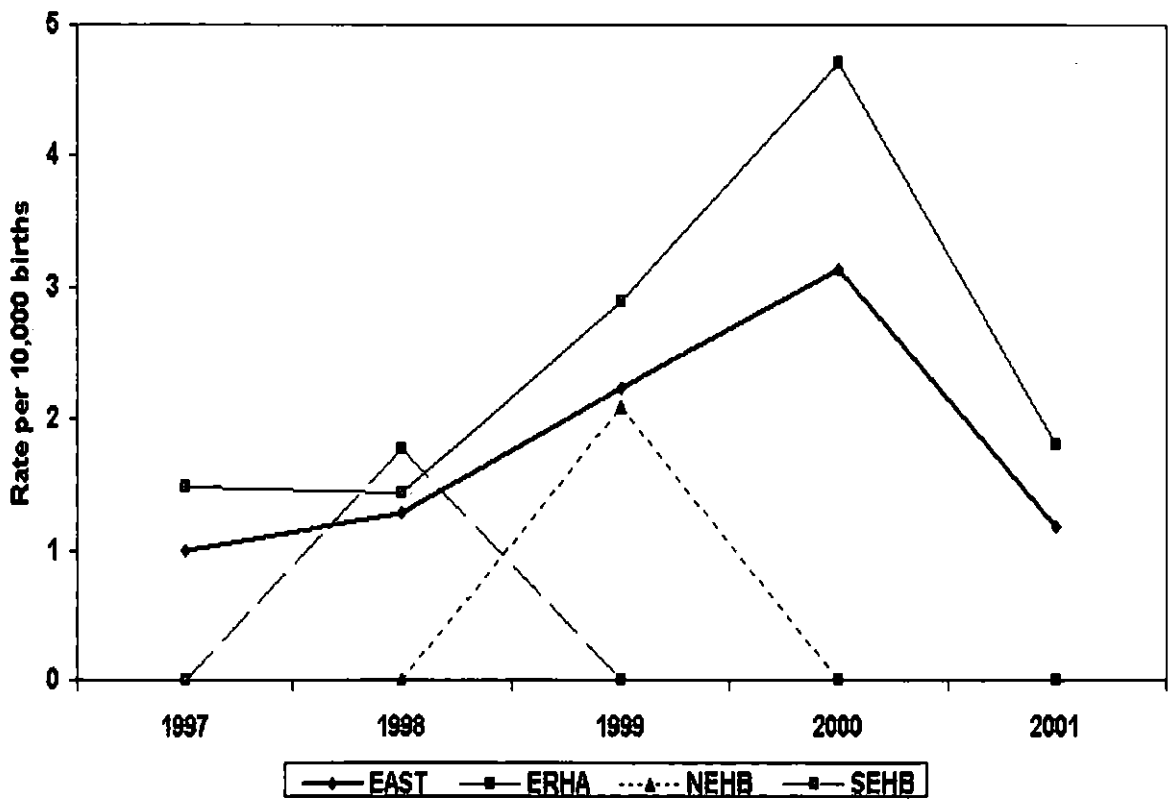
Gastroschisis

Gastroschisis is characterised by visceral herniation through the abdominal wall lateral to an intact umbilical cord and not covered by a membrane. Although the cause is generally unknown, studies have shown associations with younger mothers, inadequate nutrition, smoking and use of illicit drugs. Surgical treatment is required soon after birth. During the five-year period, 28 children were born with gastroschisis, almost all were in the catchment of the ERHA. The average birth prevalence rate per 10,000 births was 1.8 (Table 23, Fig. 22), with a continuous rise from 1997 to a peak in 2000.

Table 23. Gastroschisis: numbers and birth prevalence rates per 10,000 births 1997-2001

	EAST			ERHA			NEHB			SEHB		
YEAR	Births	No.	Rate /10,000	Births	No.	Rate /10,000	Births	No.	Rate /10,000	Births	No.	Rate /10,000
1997	**30,300**	**3**	**1.0**	20,285	3	1.5	4,532	0	0.0	5,483	0	0.0
1998	**31,092**	**4**	**1.3**	20,899	3	1.4	4,540	0	0.0	5,653	1	1.8
1999	**31,324**	**7**	**2.2**	20,767	6	2.9	4,786	1	2.1	5,771	0	0.0
2000	**31,861**	**10**	**3.1**	21,229	10	4.7	4,826	0	0.0	5,806	0	0.0
2001	**33,835**	**4**	**1.2**	22,137	4	1.8	5,378	0	0.0	6,320	0	0.0
Total	**158,412**	**28**	**1.8**	105,317	26	2.5	24,062	1	0.4	29,033	1	0.3

Fig. 22. Gastroschisis: trends in birth prevalence rates per 10,000 births 1997-2001



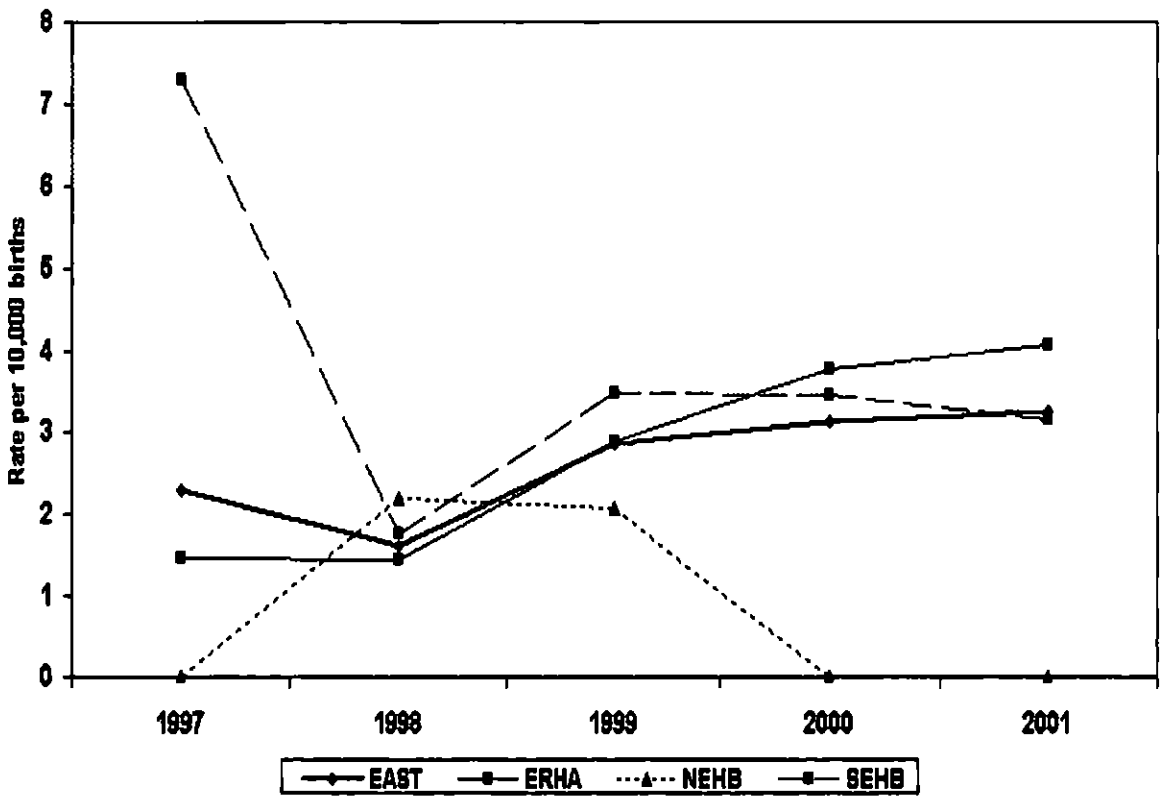
Omphalocoele

In omphalocoele, herniation of abdominal contents occurs through the umbilical insertion and covered by a membrane that may or may not be intact. The epidemiology of omphalocoele is different from gastroschisis. At least half of children with omphalocoele have additional associated anomalies such as chromosomal trisomies, cardiac or renal defects. The prognosis is mainly dependant on the presence or absence of associated anomalies. There were 42 children born with an omphalocoele during the five-year period (Table 24, Fig. 23). Three quarters were in the catchment of the ERHA and most of the remainder in the SEHB. The overall average rate per 10,000 births was 2.7 with higher rates from 1999-2001.

Table 24. Omphalocoele: numbers and birth prevalence rates per 10,000 births 1997-2001

	EAST			ERHA			NEHB			SEHB		
YEAR	Births	No.	Rate /10,000	Births	No.	Rate /10,000	Births	No.	Rate /10,000	Births	No.	Rate /10,000
1997	**30,300**	**7**	**2.3**	20,285	3	1.5	4,532	0	0.0	5,483	4	7.3
1998	**31,092**	**5**	**1.6**	20,899	3	1.4	4,540	1	2.2	5,653	1	1.8
1999	**31,324**	**9**	**2.9**	20,767	6	2.9	4,786	1	2.1	5,771	2	3.5
2000	**31,861**	**10**	**3.1**	21,229	8	3.8	4,826	0	0.0	5,806	2	3.4
2001	**33,835**	**11**	**3.3**	22,137	9	4.1	5,378	0	0.0	6,320	2	3.2
Total	**158,412**	**42**	**2.7**	105,317	29	2.8	24,062	2	0.8	29,033	11	3.8

Fig. 23 Omphalocoele: trends in birth prevalence rates per 10,000 births 1997-2001



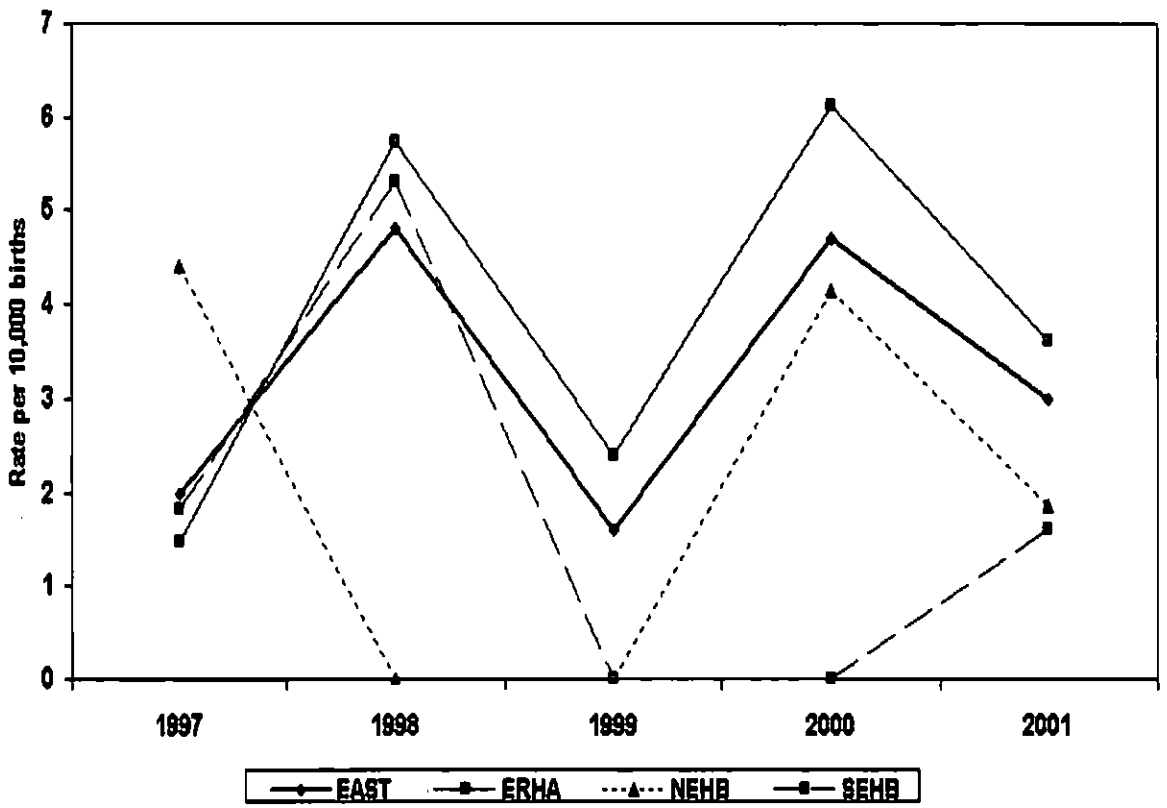
CONGENITAL ANOMALIES OF THE DIAPHRAGM

More than 90% of congenital anomalies of the diaphragm are diaphragmatic hernia. These have a high mortality and generally require immediate surgery at birth. A small proportion is associated with other anomalies. There were 51 children born with diaphragmatic anomalies during the five-year period (Table 25, Fig. 24). The overall average rate per 10,000 births was 3.2 with higher rates in 1998 and 2000, with a higher prevalence in 1998 and 2000. The majority of children were in the ERHA catchment.

Table 25. Diaphragm anomalies: numbers and birth prevalence rates per 10,000 births 1997-2001

	EAST			ERHA			NEHB			SEHB		
YEAR	Births	No.	Rate /10,000	Births	No.	Rate /10,000	Births	No.	Rate /10,000	Births	No.	Rate /10,000
1997	30,300	6	2.0	20,285	3	1.5	4,532	2	4.4	5,483	1	1.8
1998	31,092	15	4.8	20,899	12	5.7	4,540	0	0.0	5,653	3	5.3
1999	31,324	5	1.6	20,767	5	2.4	4,786	0	0.0	5,771	0	0.0
2000	31,861	15	4.7	21,229	13	6.1	4,826	2	4.1	5,806	0	0.0
2001	33,835	10	3.0	22,137	8	3.6	5,378	1	1.9	6,320	1	1.6
Total	158,412	51	3.2	105,317	41	3.9	24,062	5	2.1	29,033	5	1.7

Fig. 24. Diaphragm anomalies: trends in birth prevalence rates per 10,000 births 1997-2001



RENAL ANOMALIES

There are two main anomalies of the kidneys, renal agenesis and polycystic kidneys.

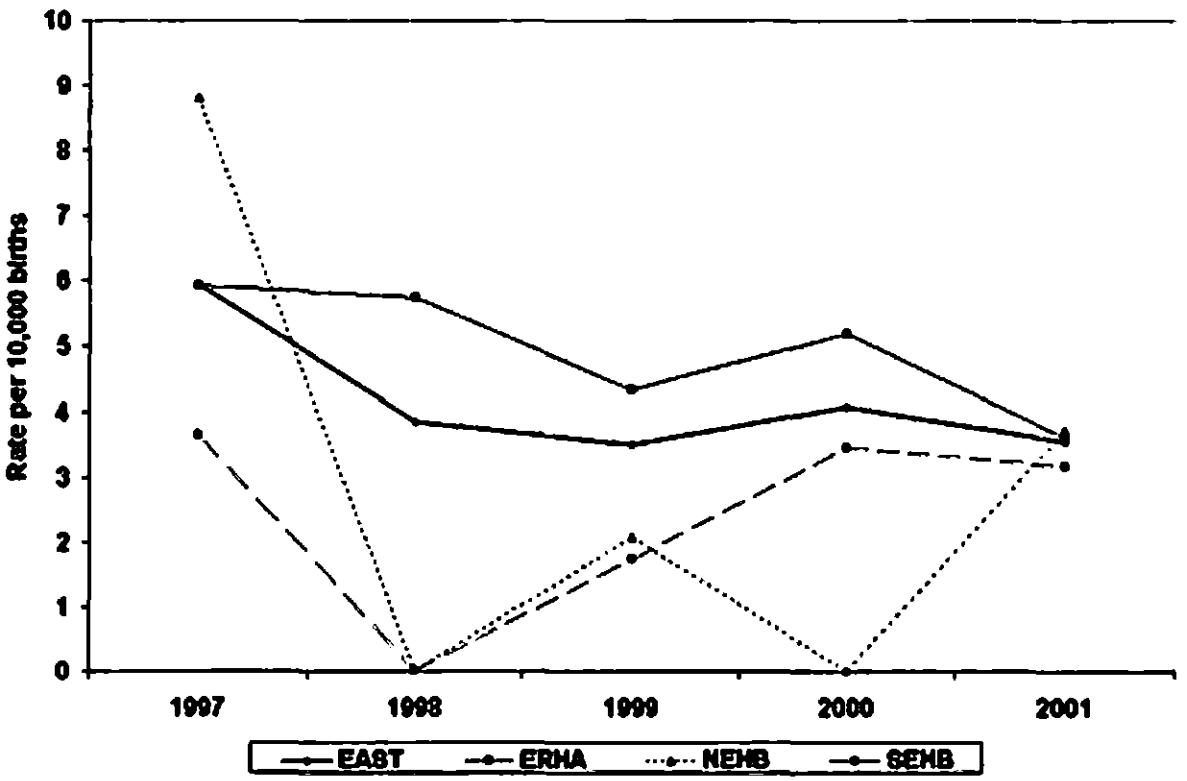
Renal agenesis

Renal agenesis is characterised by a complete absence of kidneys bilaterally or severely dysplastic kidneys. It gives rise to Potter's syndrome if both kidneys are affected which is usually lethal in the first few days of life. The cause is unclear but thought to be multi-factorial. There were 66 children born with renal agenesis during the five-year period (Table 26, Fig. 25). The overall rate was 4.2 per 10,000 births, and varied slightly from year to year within the health boards/authority.

Table 26. Renal agenesis: numbers and birth prevalence rates per 10,000 births 1997-2001

	EAST			ERHA			NEHB			SEHB		
YEAR	Births	No.	Rate /10,000	Births	No.	Rate /10,000	Births	No.	Rate /10,000	Births	No.	Rate /10,000
1997	30,300	18	5.9	20,285	12	5.9	4,532	4	8.8	5,483	2	3.6
1998	31,092	12	3.9	20,899	12	5.7	4,540	0	0.0	5,653	0	0.0
1999	31,324	11	3.5	20,767	9	4.3	4,786	1	2.1	5,771	1	1.7
2000	31,861	13	4.1	21,229	11	5.2	4,826	0	0.0	5,806	2	3.4
2001	33,835	12	3.5	22,137	8	3.6	5,378	2	3.7	6,320	2	3.2
Total	158,412	66	4.2	105,317	52	4.9	24,062	7	2.9	29,033	7	2.4

Fig. 25. Renal agenesis: trends in birth prevalence rates per 10,000 births 1997-2001



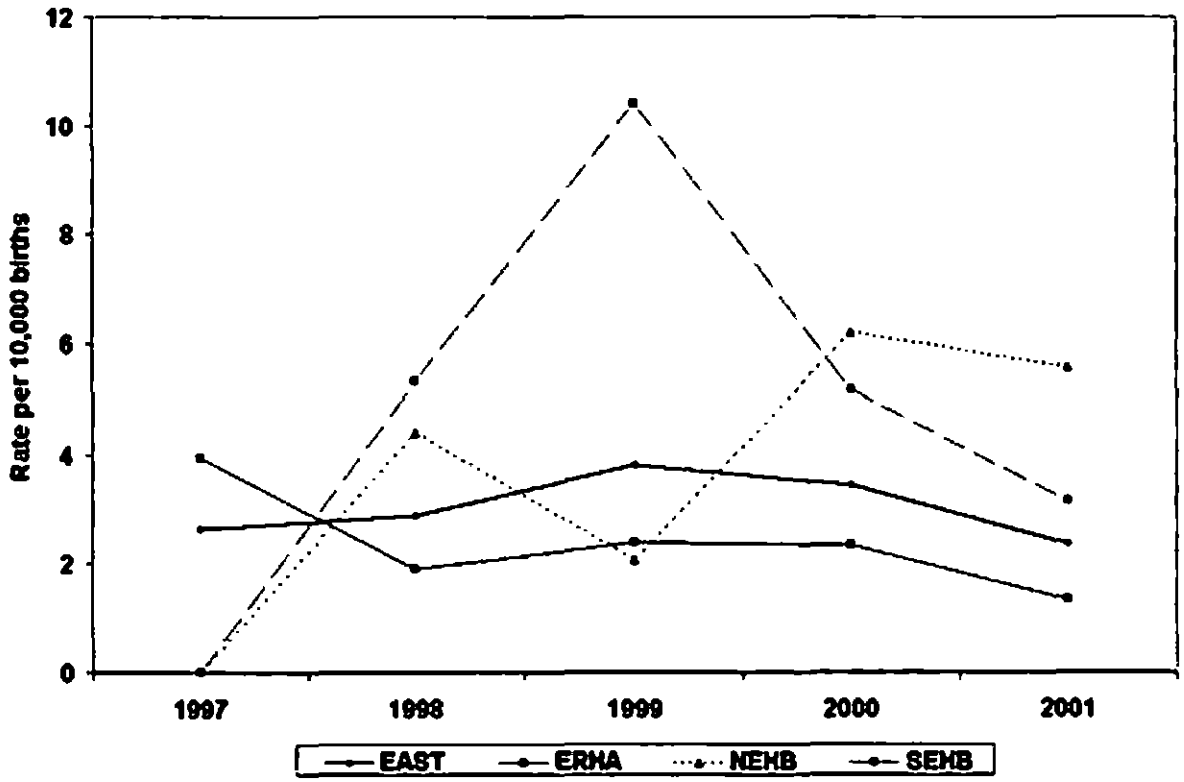
Polycystic kidneys

Cystic kidney disease is a form of renal dysplasia and is characterised by multiple cysts in the kidneys. It may be genetically inherited or associated with chromosomal or non-chromosomal syndromes, or occur sporadically. It can also result in Potter's syndrome. The prognosis depends on the extent of the anomaly and associated anomalies, ranging from having a normal life expectancy to being lethal. There were 48 children born with cystic kidney disease during the five-year period (Table 27, Fig. 26). The overall rate was 3.0 per 10,000 births, higher in 1999 and 2000. A peak was observed in the SEHB in 1999.

Table 27. Cystic kidneys: numbers and birth prevalence rates per 10,000 births 1997-2001

	EAST			ERHA			NEHB			SEHB		
YEAR	Births	No.	Rate /10,000	Births	No.	Rate /10,000	Births	No.	Rate /10,000	Births	No.	Rate /10,000
1997	**30,300**	**8**	**2.6**	20,285	8	3.9	4,532	0	0.0	5,483	0	0.0
1998	**31,092**	**9**	**2.9**	20,899	4	1.9	4,540	2	4.4	5,653	3	5.3
1999	**31,324**	**12**	**3.8**	20,767	5	2.4	4,786	1	2.1	5,771	6	10.4
2000	**31,861**	**11**	**3.5**	21,229	5	2.4	4,826	3	6.2	5,806	3	5.2
2001	**33,835**	**8**	**2.4**	22,137	3	1.4	5,378	3	5.6	6,320	2	3.2
Total	**158,412**	**48**	**3.0**	105,317	25	2.4	24,062	9	3.7	29,033	14	4.8

Fig. 26. Cystic kidneys: trends in birth prevalence rates per 10,000 births 1997-2001



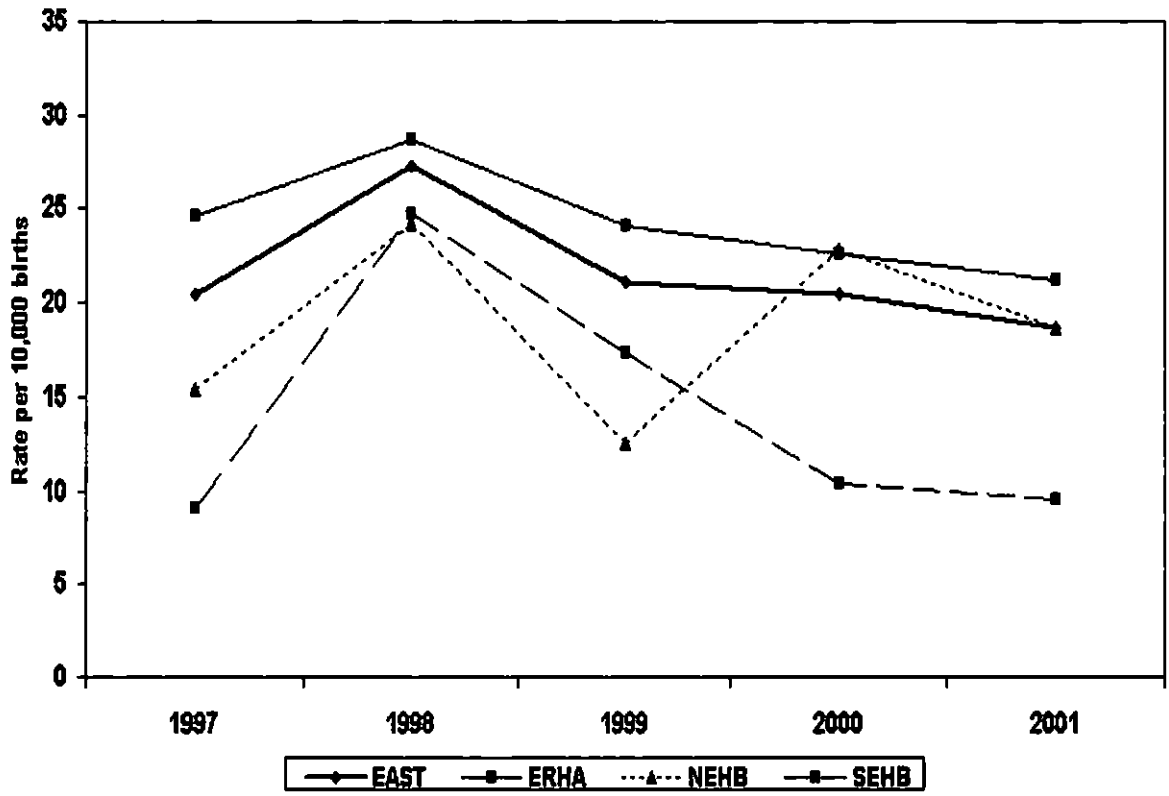
GENITAL ORGAN ANOMALIES

Genital organ anomalies include all anomalies of the male and female genital tract both external and internal. Hypospadias comprises 80% of all genital organ anomalies. Hypospadias is characterised by the opening of the urethra on the ventral side of the penis, or scrotum, or perineum. Several factors have been implicated, including genetic, endocrine and environmental influences. Treatment is through surgical repair. There were 341 children born with genital organ anomalies during the five-year period (Table 28, Fig. 27), with a higher prevalence in 1998 in the ERHA catchment and SEHB. The overall rate was 21.5 per 10,000 births, although lower in the SEHB and NEHB.

Table 28. All Genital anomalies: numbers and birth prevalence rates per 10,000 births 1997-2001

	EAST			ERHA			NEHB			SEHB		
YEAR	Births	No.	Rate /10,000	Births	No.	Rate /10,000	Births	No.	Rate /10,000	Births	No.	Rate /10,000
1997	30,300	62	20.5	20,285	50	24.6	4,532	7	15.4	5,483	5	9.1
1998	31,092	85	27.3	20,899	60	28.7	4,540	11	24.2	5,653	14	24.8
1999	31,324	66	21.1	20,767	50	24.1	4,786	6	12.5	5,771	10	17.3
2000	31,861	65	20.4	21,229	48	22.6	4,826	11	22.8	5,806	6	10.3
2001	33,835	63	18.6	22,137	47	21.2	5,378	10	18.6	6,320	6	9.5
Total	158,412	341	21.5	105,317	255	24.2	24,062	45	18.7	29,033	41	14.1

Fig. 27. All Genital anomalies: trends in birth prevalence rates per 10,000 births 1997-2001



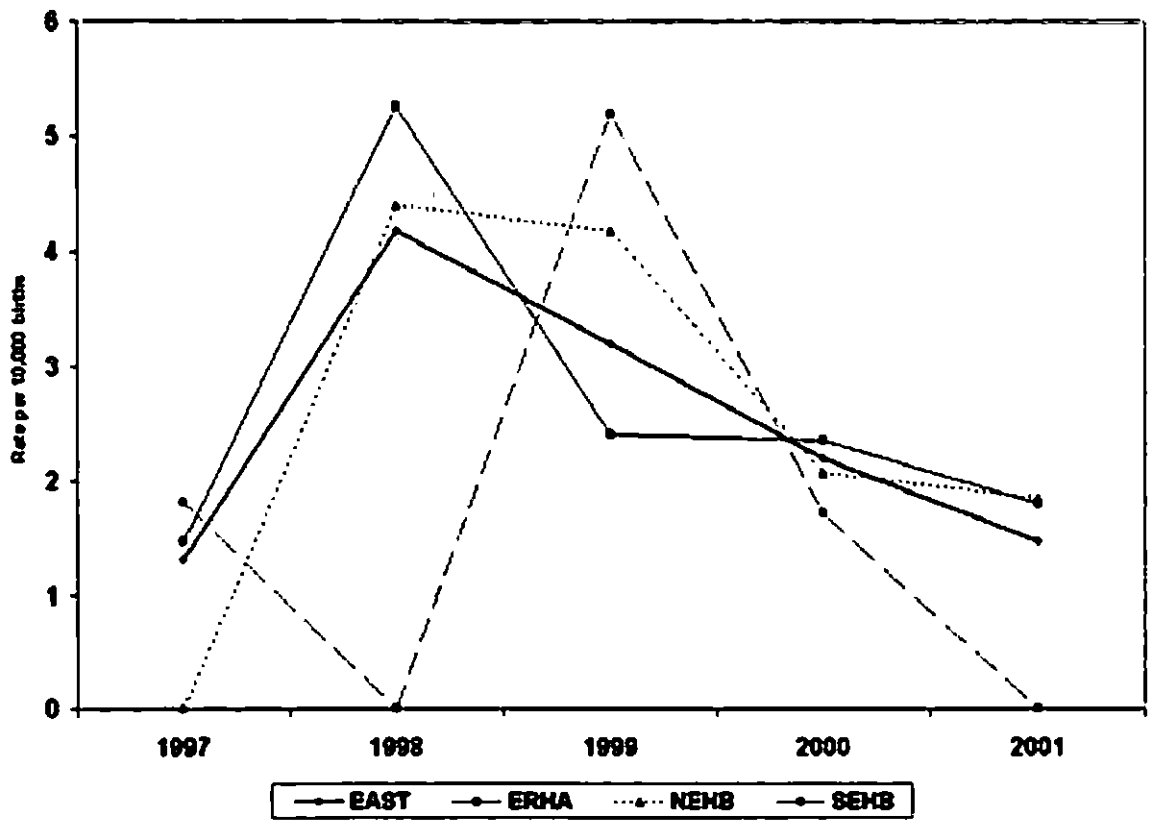
EYE ANOMALIES

Eye anomalies studied were anopthalmos and microphthalmos, which are an apparent absence and very small eyes - some normal adnexal elements and eyelids are usually present. They may be genetically inherited, or associated with chromosomal or non-chromosomal syndromes e.g. Trisomy 13. They may also be a consequence of maternal infection during pregnancy with rubella or toxoplasmosis, or the cause may be unknown. There were 39 children born with anopthalmos or microphthalmos during the five-year period (Table 29, Fig. 28), with a higher birth prevalence in 1998, particularly in the ERHA catchment. The overall rate was 2.5 per 10,000 births, although lower in the SEHB.

Table 29. Anopthalmos / microphthalmos: numbers and birth prevalence rates per 10,000 births 1997-2001

	EAST			ERHA			NEHB			SEHB		
YEAR	Births	No.	Rate /10,000	Births	No.	Rate /10,000	Births	No.	Rate /10,000	Births	No.	Rate /10,000
1997	30,300	4	1.3	20,285	3	1.5	4,532	0	0.0	5,483	1	1.8
1998	31,092	13	4.2	20,899	11	5.3	4,540	2	4.4	5,653	0	0.0
1999	31,324	10	3.2	20,767	5	2.4	4,786	2	4.2	5,771	3	5.2
2000	31,861	7	2.2	21,229	5	2.4	4,826	1	2.1	5,806	1	1.7
2001	33,835	5	1.5	22,137	4	1.8	5,378	1	1.9	6,320	0	0.0
Total	158,412	39	2.5	105,317	28	2.7	24,062	6	2.5	29,033	5	1.7

Fig 28. Anopthalmos / microphthalmos: trends in birth prevalence rate per 10,000 births 1997-2001



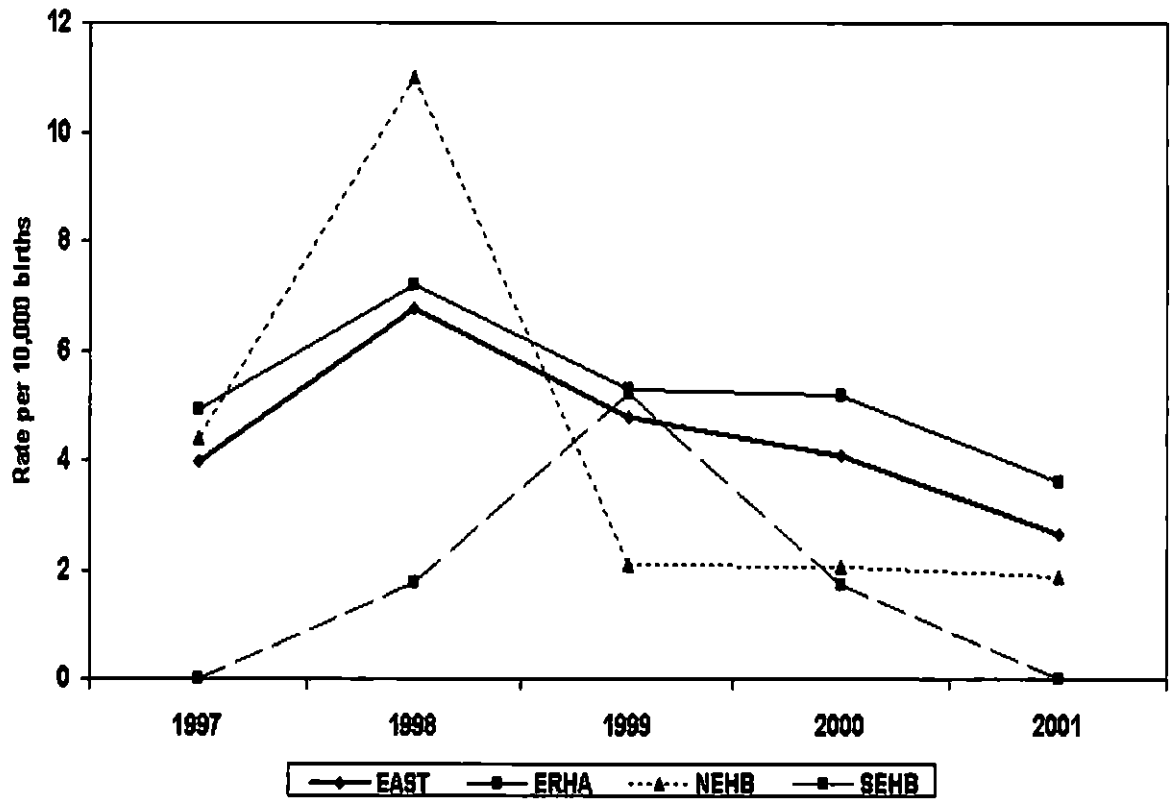
CYSTIC FIBROSIS

Cystic fibrosis is an inherited autosomal recessive disorder, and affects multiple organ systems, especially the lungs and pancreas. There were 70 children born with cystic fibrosis during the five-year period (Table 30, Fig. 29). The overall rate was 4.4 per 10,000 births, with higher rates in the NEHB and ERHA catchment in 1998. The rates in 2001 are lower because of incomplete ascertainment, due to a slightly later diagnosis in some children compared with other congenital anomalies.

Table 30. Cystic Fibrosis: numbers and birth prevalence rates per 10,000 births 1997-2001

	EAST			ERHA			NEHB			SEHB		
YEAR	Births	No.	Rate /10,000	Births	No.	Rate 10,000	Births	No.	Rate /10,000	Births	No.	Rate /10,000
1997	**30,300**	**12**	**4.0**	20,285	10	4.9	4,532	2	4.4	5,483	0	0.0
1998	**31,092**	**21**	**6.8**	20,899	15	7.2	4,540	5	11.0	5,653	1	1.8
1999	**31,324**	**15**	**4.8**	20,767	11	5.3	4,786	1	2.1	5,771	3	5.2
2000	**31,861**	**13**	**4.1**	21,229	11	5.2	4,826	1	2.1	5,806	1	1.7
2001	**33,835**	**9**	**2.7**	22,137	8	3.6	5,378	1	1.9	6,320	0	0.0
Total	**158,412**	**70**	**4.4**	105,317	55	5.2	24,062	10	4.2	29,033	5	1.7

Fig. 29. Cystic Fibrosis: trends in birth prevalence rates per 10,000 births 1997-2001



DISCUSSION

This report provides information on numbers, rates and trends in congenital anomalies in the East of Ireland. Although previous publications have provided data on congenital anomalies in some parts of the country, or on specific anomalies, this is the first report with a large geographical coverage and high proportion of births nationally.

There were 3,842 children born with congenital anomalies in the East of Ireland from 1997-2001, representing 2.4% of all births during the five-year period. The gradual fall in the number of total anomalies in 2001 is due to incomplete ascertainment that year, mostly of less serious anomalies. However, ascertainment of major anomalies is virtually complete for that year.

The birth prevalence of neural tube defects was relatively stable during the period, following a large decline that occurred during the 1980s and early 1990s. Nevertheless, 164 children were born with a NTD. Research has shown that up to 50% of NTD can be prevented through adequate dietary intake of the vitamin folic acid in the peri-conceptional period.

There was a gradual rise in the number and rate of chromosomal anomalies including Trisomy 21 and Trisomy 13 as a result of increasing maternal age in recent years. The rise in the numbers of chromosomal anomalies has not been uniform, but has shown peaks in 1998 and 2000. There was also a rise in the number of anomalies known to be associated with chromosomal anomalies such as holoprosencephaly. Although the total number and rate of congenital heart defects remained stable during the period, the rate in 1999 was higher in three of the four main types of heart defect (hypoplastic left heart, transposition of the great vessels and coarctation of the aorta), the reasons for which are unclear. The birth prevalence rate for facial cleft anomalies and digestive anomalies fluctuated from year to year, although it was not within the scope of this report to examine in more detail the individual types of facial cleft, which may have showed more pronounced trends.

All limb anomalies showed a downward trend that by 2001 could be explained by incomplete ascertainment of minor limb anomalies. Conversely, polydactyly has become more prominent in recent years with the influx of African immigrants, among whom there is a higher prevalence of polydactyly in families, probably as a result of autosomal dominant inheritance, which accounted for the steep rise in polydactyly seen in 2001. With regard to abdominal wall defects, an increasing trend was evident for both gastroschisis (1997 to 2000) and omphalocoele (1997 to 2001). The reasons for each are different. The rise in gastroschisis occurred primarily among children born to younger mothers and is possibly related to inadequate nutrition or behavioural/social habits. Omphalocoele is associated with chromosomal trisomies, which increased during the five-year period.

The rates of renal anomalies fluctuated during the five-year period. Renal agenesis and polycystic kidney disease are inter-related, in that the case definition has changed in the past decade with new technology. Some cases diagnosed as renal agenesis formerly, are known to have small cysts and are therefore polycystic kidneys, although the overall number of these anomalies would be the same. The rate for

genital anomalies remained stable throughout the period, whereas the rate for eye anomalies showed a peak in 1998 and 1999. The rate for cystic fibrosis was stable throughout the period.

This report is published at a time of major structural change within the health services, with the abolition of the previous health board structure. However, these changes are at an administrative and service provision level, while the burden of disease remains the same regardless of administrative boundaries. Thus, we have presented the data primarily as a measure of congenital anomalies at a broad geographical level - East of Ireland, while including data for each of the previous health board structures (ERHA, NEHB, SEHB) during the time of change. The data on congenital anomalies is gathered at county level and although the numbers are generally small, they do provide the opportunity for further analysis whatever administrative or service provision boundaries exist.

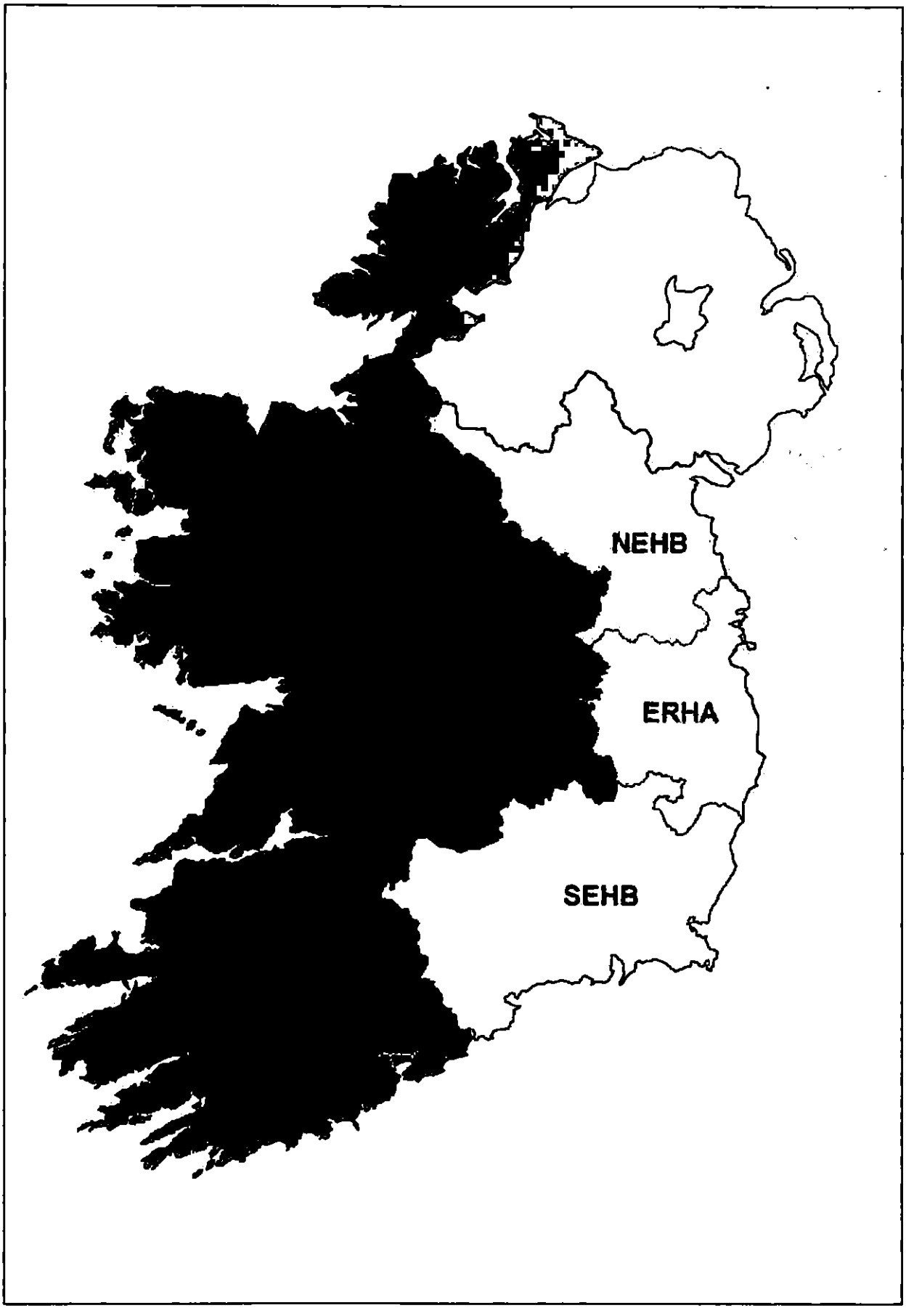
This report provides important baseline data, against which future numbers and rates of congenital anomalies in the East of Ireland or elsewhere in the country can be compared. Without this information, it would be impossible to determine if a rise in the number of children born with a particular birth defect was an expected event or something unexpected that required further investigation. It has been shown from the experience of congenital anomaly networks in Europe (EUROCAT network) and internationally (International Clearinghouse for Birth Defects Monitoring Systems – ICBD) that rising trends and clusters of individual congenital anomalies frequently occur, and that geographical and temporal comparisons using data from congenital anomaly registers is an essential component in any investigation. In recent years data from registries has been indispensable when investigating concerns regarding the rising rate of gastroschisis, chromosomal anomalies, the effects of hazardous landfill sites and unexpected disasters such as Chernobyl. Birth defect registries also provide an early-warning system and limit the consequences of disasters such as occurred with the drug thalidomide in the 1960s.

National surveillance of congenital anomalies is lacking in Ireland, unlike other European countries such as Norway, Finland, England, Wales and Sweden. This activity is relatively inexpensive to undertake and maintain. It is also much more cost-effective and timely than initiating ad-hoc surveys to investigate a rise in a specific birth defect in a particular region, where there is no congenital anomaly surveillance. Congenital anomalies often have life-long consequences for both the individual and the health and social services as well as the fact that some are preventable e.g. neural tube defects, congenital rubella syndrome. The effectiveness of preventive measures can be assessed and monitored using data from congenital anomaly registers.

During the writing of this report, the NEHB registry ceased surveillance at the end of 2001, however, it is hoped that data collection will recommence soon. Overall from 2002, there has been no congenital anomalies surveillance for almost one third of births in the country, due to an absence of registries in some of the health boards. This means that any apparent rise in birth defects in those areas that gives rise to concern will need a special study simply to find out what the baseline is, before any further investigation. Extending congenital anomaly surveillance nationally would be a prudent step in the context and framework of a modern health service.

APPENDIX 1

Health Board Catchments 1997-2001



APPENDIX 2

Congenital Anomaly Subgroups: ICD 9 and ICD 10 Codes

Nervous System	740,741, 7420-7425, 7428, 7429	Q00, Q01, Q02,Q03,Q04,Q05, Q06, Q07
Neural Tube Defects:	7400-7420,	Q00, Q01, Q05
Anencephalus	7400-7402	Q00
Encephalocoele	7420	Q01
Spina Bifida	7410-7419	Q05
Eye		
Anophthalmos/microphthalmos	7430, 7431	Q110, Q111, Q112
Congenital Heart Disease		
Anomalies of cardiac chambers and connections	74500, 7451, 7453, 7457	Q20
Common arterial truncus	74500	Q200
Transpositions of great vessels, complete Single ventricle	74510, 7453	Q203, Q204
Tetralogy of Fallot	7452	Q213
Hypoplastic left heart	7467	
Coarctation of aorta	7471	Q251
Cleft Lip With or Without Cleft Palate	7491, 7492	Q36-Q37
Digestive System	7503-7505, 7507-7519	Q39, Q400, Q402
Internal Urogenital System:		
Bilateral renal agenesis	75300	Q601, Q604, Q606
Cystic kidney disease	7531	Q61
External genital system		
Hypospadias	75260	Q541-Q543, Q548
Limb	7543-7544[excl 75432] 7545-7547 [excl 75452, 75460, 75473], 7550-7551, 7552-7554, 7555-7556 [excl 75560, 7558-7559	Q650-Q656, Q66[excl Q662, Q664, Q668], Q682-Q685, Q69, Q71, Q72, Q73, Q74
Limb reduction	7552-7554	Q71- Q73
Polydactyly	7550	Q69
Musculoskeletal &Connective Tissue		
Diaphragmatic hernia	75661	Q790
Omphalocele	75670	Q792
Gastroschisis	75671	Q793
Chromosomal	7580-7583, 7585-7589 [excl 758620]	Q90-Q94, Q96-Q99
Down syndrome	7580	Q90
Patau syndrome/trisomy 13	7581	Q914-Q917
Edwards syndrome/trisomy 18	7582	Q910-Q913

APPENDIX 3

STAFF OF THE REGISTRIES

The staff of the registries involved in this study are as follows:

Eastern Regional Health Authority Registry (Dublin EUROCAT Registry)

Dr. Bob Mc Donnell, Specialist in Public Health Medicine.

Ms. Virginia Delany, Research Nurse.

Dr Howard Johnson, Specialist in Public Health Medicine *.

Dr Zachary Johnson, Specialist in Public Health Medicine * RIP.

South Eastern Health Board Registry

Dr. BethAnn Roch, Specialist in Public Health Medicine.

Ms. Johanna Costigan, Research Nurse.

Dr. Maire O'Connor Specialist in Public Health Medicine *.

Ms. Brenda Cooper, Research Nurse *.

North Eastern Health Board Registry

Dr. Fenton Howell, Specialist in Public Health Medicine.

Ms. Deirdre Mulligan, Research Nurse.

Note: * former staff working in the registry during the study period

APPENDIX 4

Counties covered by the registries with annual number of live births 1997-2001

COUNTY	1997	1998	1999	2000	2001
DUBLIN	16,085	16,532	16,288	16,392	17,051
WICKLOW	1,601	1,682	1,783	1,824	1,912
KILDARE	2,470	2,571	2,570	2,881	3,050
CARLOW	547	663	620	706	731
WEXFORD	1,574	1,638	1,707	1,616	1,769
KILKENNY	989	1,022	983	1,047	1,115
TIPPERARY SOUTH	984	914	1,044	987	1,080
WATERFORD	1,356	1,355	1,389	1,423	1,584
CAVAN	793	729	781	714	756
MONAGHAN	636	657	690	627	626
LOUTH	1,370	1,427	1,471	1,535	1,728
MEATH	1,699	1,693	1,816	1,921	2,235
TOTAL	**30,104**	**30,883**	**31,142**	**31,673**	**33,637**

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Title: Congenital Anomalies in the East of Ireland 1997 - 2001
Published by: HSE Eastern Region 2005
Design and output: Accent Publications Limited
Printed by: Ebrook

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