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Stability of prevalence rates of anorectal malformations in the Alberta Congenital Anomalies Surveillance System 1990-2004

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Abstract

Background/Purpose: Anorectal malformations appeared to be increasing in the province of Alberta, Canada. To assess whether this was a significant trend, with the possibility of these having a teratogenic origin, we examined the frequency of anorectal malformations over a 15-year period.

Methods: We examined the records of the Alberta Congenital Anomaly Surveillance System, which is a semiactive surveillance system using the British Paediatric Association and the Royal College of Paediatrics and Child Health expansions of the *International Classification of Diseases–Ninth Revision* and the *International Classification of Diseases–10th Revision*.

Results: The overall rate was 1/2162 (4.63/10,000 total births) with a marked male predominance (1.7:1). Approximately two thirds of the 273 cases had 1 or more malformations.

Conclusion: Although there was an increasing trend in the rate from 1999 especially for the multiples, this was not significant. In view of the advances in syndrome identification and molecular diagnostics, consideration should be given to a detailed review of the family history and appropriate testing not only for multiple cases but also for isolated ones.

The year 1999 saw the beginning of an upward trend in the frequency of anorectal malformations (ARMs) both in the Canadian national data (1997—4.61/10,000 births; 1998—5.18; 1999—6.06) and in our provincial data. A detailed survey of the Alberta data was undertaken because of our ability to obtain further details on individual cases.

1. Methods

Congenital anomaly reporting forms are the main source of information for the Alberta Congenital Anomaly Surveillance System (ACASS), and these were examined for the British Paediatric Association (BPA) and the Royal College of Paediatrics and Child Health (RCPCH) adaptations of the *International Classification of Diseases–Ninth Revision* (*ICD-9*) for the rubrics 751.21 to 751.24 and the *International Classification of Diseases–10th Revision* (*ICD-10*) Q42.0 to Q42.3 (see Appendix). Terminations of pregnancy are not included in the overall rates, although

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Table 1 Birth prevalence and CIs for anorectal malformations in Alberta 1990-2004 (BPA adaptation of *ICD-9* 751.21, 751.22, 751.23, 751.24; RCPCH adaptation of *ICD-10* Q42.0, Q42.1, Q42.2, Q42.3)

	n	Total births	Rate/10,000	95% CI	Frequency
Total	273	590081	4.63	4.11-5.21	1/2162
Male	170	302408	5.62	4.81-6.54	1/1779
Female	100	287626	3.48	2.83-4.23	1/2876
Indeterminate	3	47	(1.1% of all ARMs were indeterminate)		

they have been collected by the ACASS since 1997 and have been evaluated separately. We have divided our caseload into (*a*) isolated ARMs and (*b*) ARMs with additional malformations. The latter group is subdivided into chromosomal conditions, syndromes of known origin, syndromes of uncertain origin, associations, and sequences, with the remainder categorized as multiple congenital anomalies (MCAs). A special section has been devoted to the VATER/VACTERL (V-V) association. Although the ACASS is not an active surveillance system, nevertheless because of its multiple sources of ascertainment and the ability to verify diagnoses from clinical records, physicians' reports, x-rays, laboratory records, autopsies, and liaison with vital statistics (birth, death, and stillbirth records), we believe we have close to full ascertainment for this malformation [1].

2. Results

The overall rate for ARMs for Alberta for the years 1990-2004 is seen in Table 1 and shows a marked male preponderance (1.7:1). There were 82 isolated cases (51 males, 31 females) and 191 cases with multiple anomalies (119 males, 69 females, 3 indeterminate) giving prevalence rates of 1.39 (1/7196) and 3.24 (1/3089) per 10,000 total births for isolated and multiple anomaly cases, respectively. Males comprise 62% of cases in both the isolated and multiple anomaly cases. Table 2 shows the annual rates and confidence intervals (CIs) for the 15 years, and the average birth weight, gestational age, and maternal ages for the 3 categories are seen in Table 3.

Information on fistulas is probably underreported because there were only 88 (32%) in 273 of cases with a fistula; 22 (25%) of the 88 had an isolated ARM. Thirteen of 82 (16%) isolated cases were described as having stenosis, 2 had a membrane, and 1 was ectopic.

Table 2 Annual birth prevalence rates and CIs for ARMs, 1990-2004 (ACASS data)

Year	n	Rate/10,000	95% CI	Frequency
1990	24	5.60	3.59-8.32	1/1787
1991	21	4.92	3.05-7.52	1/2032
1992	15	3.58	2.01-5.89	1/2796
1993	20	4.98	3.05-7.69	1/2008
1994	22	5.54	3.48-8.38	1/1806
1995	18	4.64	2.76-7.33	1/2155
1996	15	3.98	2.23-6.55	1/2514
1997	14	3.81	2.08-6.37	1/2628
1998	11	2.91	1.46-5.18	1/3437
1999	16	4.21	2.41-6.82	1/2377
2000	16	4.34	2.49-7.04	1/2302
2001	20	5.34	3.27-8.24	1/1873
2002	22	5.71	3.58-8.64	1/1752
2003	23	5.73	3.64-8.59	1/1745
2004*	16	3.94	2.26-6.39	1/2537
1990-2004	273	4.63	4.10-5.21	1/2162

* Ascertainment may be incomplete.

2.1. Anorectal malformations with multiple anomalies

The most frequent entity belonging in this category was that of the V-V association. We used a broad definition and had 35 cases (Table 4). Those cases with chromosomal syndromes are seen in Table 5, and the remaining syndromes and sequences are in Table 6.

2.2. Other MCAs with ARMs

The 91 cases designated as MCAs had a large number of different combinations that did not fit into any designated syndrome, association, or sequence. Many of them had components of V-V but had so many other anomalies that it was decided to classify them as MCAs. The most common associated systems were urinary/renal, genital, musculoskel-

Table 3 Average birth weight, gestational age, and maternal age for ARMs in Alberta, 1990-2004

	Mean	SD	Range
Birth weight (g)			
Isolated	3319.63	549.87	1847-4590
Multiple[a]	2333.06	1122.85	120-4510
VATER	2681.31	851.30	780-4510
Gestational age (completed wk)			
Isolated	38.79	1.70	32-42
Multiple[a]	34.54	6.15	20-42
VATER	36.86	3.69	24-42
Maternal age (y)			
Isolated	28.15	6.64	16-45
Multiple[a]	29.14	5.92	16-44
VATER	29.29	6.80	16-41

[a] Includes VATER as well, that is, *all* multiples.