

Web Material

Mortality and Cancer Incidence in Carriers of Balanced Robertsonian Translocations: A National Cohort Study

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Web Table 1: Cause-specific mortality in subjects cytogenetically diagnosed with balanced Robertsonian translocations during 1962-2006 in Great Britain, for translocations involving chromosomes 13 and 14 and chromosomes 14 and 21

ICD-9	Cause	Chromosomes involved							
		rob(13;14)				rob(14;21)			
		No.	SMR	95% CI	AER	No.	SMR	95% CI	AER
140-208	All malignant neoplasms	55	0.97	0.73, 1.27	-4.80	14	0.74	0.40, 1.24	-52.2
240-279	Endocrine, nutritional, metabolic, immunity	2	0.77	0.09, 2.78	-2.00	1	1.15	0.03, 6.41	1.40
290-319	Mental disorders	3	0.87	0.18, 2.55	-1.50	1	0.83	0.02, 4.60	-2.20
320-389	Diseases of the nervous system	2	0.43	0.05, 1.55	-8.90	6	3.68 ^a	1.35, 8.01	46.3
390-459	Diseases of the circulatory system	54	0.90	0.68, 1.17	-20.3	14	0.75	0.41, 1.26	-48.9
460-519	Diseases of the respiratory system	17	0.93	0.54, 1.49	-4.10	6	0.99	0.36, 2.15	-0.70
520-579	Diseases of the digestive system	7	0.78	0.31, 1.60	-6.70	4	1.38	0.38, 3.54	11.7
580-629	Diseases of the genitourinary system	2	0.96	0.12, 3.46	-0.30	0	0.0	0.0, 5.38	-7.30
710-739	Musculoskeletal system and connective tissue	0	0.0	0.0, 3.77	-3.30	1	2.81	0.07, 15.6	6.82
740-759	Congenital anomalies	2	3.12	0.38, 11.3	4.50	1	4.79	0.12, 26.5	8.40
800-999	Accidents and violence	11	1.18	0.59, 2.11	5.60	2	0.74	0.09, 2.65	-7.70
001-999	All causes ^b	160	0.93	0.79, 1.09	-40.5	50	0.90	0.67, 1.18	-60.8

Abbreviations: AER, absolute excess rate, number of excess deaths per 100,000 population per annum; CI, confidence interval; ICD, International Statistical Classification of diseases, 9th revision (1); SMR, standardised mortality ratio

a) two-sided P-value based on exact method $p < 0.05$

b) The causes of five deaths in subjects diagnosed with rob(13;14) are not listed separately (2 not known, 1 senility, 1 myelodysplastic syndrome, 1 infarction of spleen)

Web Table 2: Cancer incidence in subjects cytogenetically diagnosed with balanced Robertsonian translocations during 1962-2006 in Great Britain, for translocations involving chromosomes 13 and 14 and chromosomes 14 and 21

ICD-9 code	Cancer site	rob(13;14)			rob(14;21)		
		No.	SIR	95% CI	No.	SIR	95% CI
140-171, 173-208	All malignant neoplasms ^a	131	1.10	0.92, 1.31	39	0.98	0.70, 1.34
141-149	Tongue, mouth, pharynx	4	1.51	0.41, 3.88	1	1.22	0.03, 6.77
150	Oesophagus	2	0.76	0.09, 2.74	1	1.14	0.03, 6.34
151	Stomach	5	1.64	0.53, 3.82	1	1.01	0.03, 5.63
153, 154	Colon and rectum	6	0.45 ^b	0.17, 0.99	6	1.34	0.49, 2.91
155	Liver	1	0.85	0.02, 4.76	0	0.0	0.00, 9.45
157	Pancreas	1	0.37	0.01, 2.08	1	1.09	0.03, 6.05
162	Lung	15	0.93	0.52, 1.54	4	0.78	0.21, 1.98
163	Pleura	1	1.62	0.04, 9.00	0	0.0	0.0, 16.9
170	Bone	1	4.43	0.11, 24.7	0	0.0	0.0, 52.3
172	Cutaneous melanoma	4	0.88	0.24, 2.24	3	1.98	0.41, 5.77
174, 175	Breast	39	1.58 ^c	1.12, 2.15	6	0.73	0.27, 1.59
179,182	Corpus uteri	4	1.21	0.33, 3.09	0	0.0	0.0, 3.21
180	Cervix	2	0.89	0.11, 3.23	0	0.0	0.0, 4.94
183	Ovary	3	0.87	0.18, 2.54	1	0.83	0.02, 4.65
184.0-184.4	Vagina & vulva	1	1.76	0.04, 9.80	0	0.0	0.0, 18.8
185	Prostate	14	1.47	0.80, 2.47	4	1.18	0.32, 3.02
186	Testis	0	0.0	0.0, 3.75	1	3.77	0.10, 21.0
187.1-187.4	Penis	1	5.73	0.14, 31.9	1	18.4	0.47, 103
188	Bladder and urethra	3	0.83	0.17, 2.44	0	0.0	0.0, 3.08
189	Kidney and ureter	0	0.00	0.00, 1.23	0	0.0	0.0, 3.65
190	Eye	0	0.00	0.00, 14.7	1	12.6	0.32, 69.4
191, 192, 225, 237.5, 237.6, 237.9, 239.6	Nervous system tumours, including benign ^b	6	1.78	0.65, 3.87	2	1.78	0.22, 6.45
193	Thyroid	0	0.0	0.0, 3.35	1	2.71	0.07, 15.1
196.0-199.1	Unknown primary site	3	0.79	0.16, 2.31	1	0.77	0.02, 4.27
200, 202	Non-Hodgkin lymphoma	9	2.12	0.97, 4.03	3	2.10	0.43, 6.14
201	Hodgkin disease	0	0.00	0.00, 4.66	1	3.96	0.10, 22.1
203	Myeloma	4	2.87	0.78, 7.34	0	0.0	0.0, 7.60
204-208	Leukaemia	4	1.51	0.41, 3.86	1	1.12	0.03, 6.27

Abbreviations: CI, confidence interval; ICD, International Statistical Classification of Diseases, 9th revision (1); SIR, standardised incidence ratio

- a) All malignant neoplasms except non-melanoma skin cancer. 1 cancer (maxillary sinus) is not listed separately under the rob(13;14).
- b) two-sided P-value based on exact method $p < 0.05$
- c) two-sided P-value based on exact method $p < 0.01$
- d) The 8 observed tumours included comprise 4 malignant cancers of the brain and 4 benign neoplasms of the meninges. For England and Wales, benign nervous system neoplasms were included from the year 1971 for Scotland, from the year 2000.

Web Table 3: Incidence of cancer overall and of haematological malignancies in subjects cytogenetically diagnosed with balanced Robertsonian translocations during 1962-2006 in Great Britain, by age at cytogenetic diagnosis

ICD-9 code	Cause	Age at cytogenetic diagnosis (years)								
		0-14			15-44			45-84		
		No.	SIR	95% CI	No.	SIR	95% CI	No.	SIR	95% CI
140-171, 173-200, 202	All malignant neoplasms ^a	5	1.07	0.35, 2.50	104	1.09	0.89, 1.32	93	1.01	0.82, 1.24
201	Non-Hodgkin lymphoma	1	4.24	0.11, 23.6	7	1.93	0.78, 3.98	5	1.66	0.54, 3.88
203	Hodgkin disease	1	4.51	0.11, 25.1	0	0.0	0.0, 4.43	0	0.0	0.0, 17.2
204-208	Myeloma	0	0.0	0.0, 162	3	3.16	0.65, 9.23	1	0.76	0.02, 4.25
	Leukaemia	2	6.99	0.85, 25.2	3	1.53	0.32, 4.47	1	0.48	0.01, 2.69

Abbreviations: CI, confidence interval; ICD, International Statistical Classification of diseases, 9th revision (1); SIR, standardised incidence ratio

a) All malignant neoplasms except non-melanoma skin cancer

References

1. WHO. *International Statistical Classification of diseases and related health problems, 9th revision*. Geneva: World Health Organization; 1977.