WEST MIDLANDS CONGENITAL ANOMALY REGISTER
DOWNS SYNDROME 2002

This brief report details Down's Syndrome cases delivered during 2002 to West Midlands residents. It is a supplement to the report on Down's Syndrome 1995-1997[1] which contained more detailed analyses of cases. For the first time additional information is available on screening tests and results, collected by the network of Antenatal Screening Co-ordinators across West Midlands maternity units.

Cases are reported by place of residence at delivery in order to overcome biases introduced by referral patterns between hospitals. Cases of fetal loss before 24 weeks gestation and terminations of pregnancy are included so that incidence rates are not subject to variation in prenatal detection and termination rates.

Incidence - The incidence of Down's Syndrome during 2002 in the West Midlands was 19.8/10,000 births (1:504). After natural losses and terminations, the incidence of Down's Syndrome was 10.6/10,000 registerable births (1:945).

Detection - Prenatal diagnoses were made in 50% of Down's Syndrome cases.

Screening - Screening was carried out in 54% of Down's Syndrome cases.

Outcomes - For cases of Down's Syndrome that were live born (excluding terminations), the infant mortality rate was 36/1,000.

INCIDENCE

There were 123 cases of Down's Syndrome born to West Midlands residents during 2002. Of these, 121 were in singleton pregnancies and 2 in twin pregnancies. In both twin pregnancies, there was only one affected twin.

Table 1 - Down's Syndrome: cases by Health Authority, West Midlands 2002

Strategic Health Authority	cases	births	rate	95%CI	risk
Shropshire & Staffordshire	35	15,539	22.5	15.1 - 30.0	1: 444
Birmingham & the Black Country	53	30,213	17.5	12.8 - 22.3	1: 570
Coventry, Warks, Herefordshire & Worcs	35	16,249	21.5	14.4 - 28.7	1: 464
West Midlands	**123**	**62,001**	**19.8**	**16.3 - 23.3**	**1: 504**

Rate: per 10,000 births
95%CI: 95% confidence interval

Table 2 - Down's Syndrome: registerable cases by Health Authority, West Midlands 2002

Strategic Health Authority	cases	births	rate	95%CI	risk
Shropshire & Staffordshire	13	15,387	8.4	3.9 - 13.0	1: 1,184
Birmingham & the Black Country	35	29,934	11.7	7.8 - 15.6	1: 855
Coventry, Warks, Herefordshire & Worcs	17	16,097	10.6	5.5 - 15.6	1: 947
West Midlands	**65**	**61,418**	**10.6**	**8.0 - 13.2**	**1: 945**

Rate: per 10,000 births
95%CI: 95% confidence interval

Table 1 shows the incidence of all Down's Syndrome cases (including fetal losses and terminations). Table 2 shows the incidence of Down's Syndrome for registerable births only (i.e. live births and stillbirths); this rate is subject to variations in detection rates and termination practices.

[1] http://www.wmpi.net/car/downs/index_downs.htm

MATERNAL AGE

Table 3 - Down's Syndrome: cases by maternal age, West Midlands 2002

Age (years)	cases	births	rate	risk
<20	8	5,273	15.2	1: 659
20-24	14	13,371	10.5	1: 955
25-29	10	16,291	6.1	1: 1,629
30-34	27	17,630	15.3	1: 653
35-39	37	7,912	46.8	1: 214
40-44	25	1,451	172.3	1: 58
45+	2	73	274.0	1: 37
Total	**123**	**62,001**	**19.8**	**1: 504**

Rate: per 10,000 births
Age: maternal age at expected date of delivery

The incidence of Down's Syndrome is known to be affected by maternal age, with an increasing risk of an affected pregnancy with increasing maternal age. There is currently a general trend for women to have babies at an older age.

Table 3 shows that 76% all of babies are born to women aged between 20 and 34 years. In the West Midlands 15% of babies were born to women aged 35 years or more, whereas 52% of Down's Syndrome pregnancies occurred in women in this age group. The over 40 year old age group comprises less than 3% of the maternal population, within which 22% of the Down's Syndrome cases occurred.

Figure 1 - Down's Syndrome: cases by maternal age, West Midlands 2002

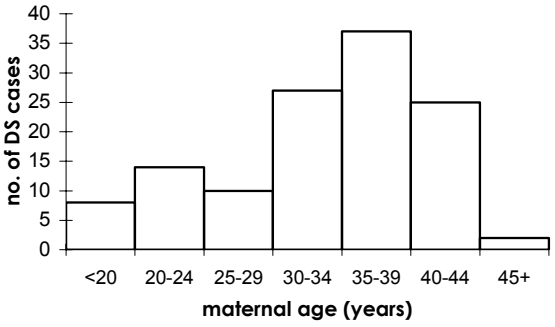

Figure 2 - Down's Syndrome: incidence by maternal age, West Midlands 2002

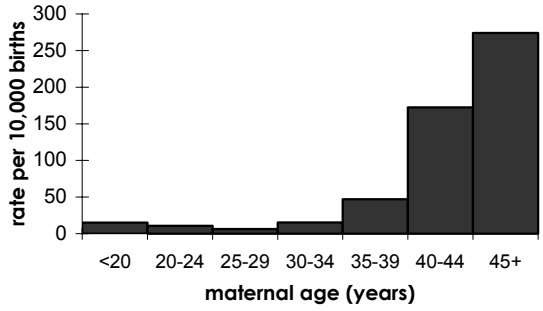

Figure 1 shows the actual number of Down's Syndrome cases in each maternal age group with the majority of cases born to women in their thirties, in contrast, Figure 2 shows the increasing incidence in older mothers. This risk is less than 1 in 900 for women aged less than 35 years and increases steeply after the age of 35.

ANTENATAL SCREENING AND PRENATAL DIAGNOSIS

Table 4 - Down's Syndrome: screening and prenatal diagnosis, West Midlands 2002

	Maternal age < 35 yrs		Maternal age >= 35 yrs		
Screening	**Prenatal Diagnosis**	**No Prenatal Diagnosis**	**Prenatal Diagnosis**	**No Prenatal Diagnosis**	**Total**
Not offered					
parents requested karyotype			7		**7**
karyotype following ultrasound			4		**4**
other (late booker/twin/IUD/TOP)	2	5	1	10	**18**
Offered and accepted					
1st trimester - screen +ve	2		2		**4**
2nd trimester - screen +ve	13	5	19	1	**38**
2nd trimester - screen -ve		19	2	3	**24**
Offered and declined					
Screening declined	1	12	8	7	**28**
Total	**18**	**41**	**43**	**21**	**123**

Prenatal diagnoses are classified as those cases in which a karyotype was established before delivery. Sixty one of 123 (50%) Down's Syndrome cases were diagnosed prenatally. All prenatal diagnoses were made before 24 weeks gestation.

Twenty eight cases (23%) were not offered screening. In seven cases the diagnosis was made as a result of karyotyping for maternal age with no other risk factors. Four cases were offered karyotyping following the detection of fetal anomalies on their booking scan. Seven cases were diagnosed as intrauterine deaths at presentation or were terminated following structural anomalies, with a postnatal karyotype. Nine cases presented too late for serum screening, of which two cases were subsequently diagnosed prenatally following abnormal ultrasound findings of structural anomalies.

Screening was carried out in 66 cases (53%). Four cases had first trimester screening, 62 had second trimester screening.

INVASIVE TESTING

Table 5 - Down's Syndrome cases: invasive testing by maternal age, West Midlands 2002

Prenatal cytogenetics	under 35 yrs	35 yrs and over	All ages
Chorionic villus sampling	3	8	11
Amniocentesis	14	34	48
Fetal blood sample +/- amnio	1	1	2
None	41	21	62
Total	**59**	**64**	**123**

Table 5 illustrates the methods of prenatal karyotyping. Chorionic villus sampling was used to make the diagnosis in 13% (8/64) of cases in the older mothers, compared to 5% (3/59) in those less than 35 years of age. Amniocentesis is the most common method of prenatal karyotyping (82%).

OUTCOMES

The outcome of affected pregnancies is dependent on both prenatal diagnosis and the presence of additional major structural anomalies.

Table 6 - Down's Syndrome: outcomes of cases, West Midlands 2002

Outcome	Total
Late fetal loss	7
Late fetal loss - TOP	51
Stillbirth	2
Stillbirth - TOP	3
Neonatal death	0
Neonatal death - TOP	4
Post neonatal death	2
Alive	54
Total	**123**

TOP: termination of pregnancy

Forty seven percent (58/123) of Down's Syndrome cases were terminated. There were 60 live births, of which six died in the first year of life - four were neonatal deaths following termination. The infant mortality rate was 100/1,000 live births (6/60). Excluding termination of pregnancy the infant mortality rate was 36/1,000 live births (2/56).

When a prenatal diagnosis was made, 93% (57/61) of cases were terminated. One case was terminated without a diagnosis of Down's Syndrome because of the presence of another structural malformation.

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