

## Objectives

The National Down Syndrome Cytogenetic Register collects all reports of trisomies 21, 18, and 13 and their genetic variants that occur in England and Wales. The NHS Fetal Anomaly Screening Programme annual report focuses the commitment to a uniform screening based on uptake and results in best practice methods within England. This report compares the results of this national register and screening program report with our own district general hospital results implementing the same screening methods and standards of care.

## Methods

The uptake, screening, and diagnostic results of all women booked from April 1, 2009 to March 31, 2013 were acquired. A retrospective study was completed on all women who screened, and the uptake was compared to the Fetal Anomaly Screening Programme. Uptake of screening was calculated by number of tests divided by number of births. Further evaluation was performed on those that had positive results and an elevated risk during the first or second trimester screenings. Evaluation of the diagnostic tests and pregnancy outcomes were investigated and compared to the National Down Syndrome Cytogenetic Register for England and Wales.

## Results

At Ealing Hospital, 14,179 women booked between April 1, 2009, and March 31, 2013. The screening test breakdown is as follows 7,709 (54%) had combined test, 1,360 (9.5%) had second trimester tests (triple or quadruple test.) From the combined test, 215 (2.7%) were considered high risk and from the second trimester test, 69 (5.1%) were considered high risk. Diagnostic testing was offered to patients who screened and had an increased risk for Downs syndrome.

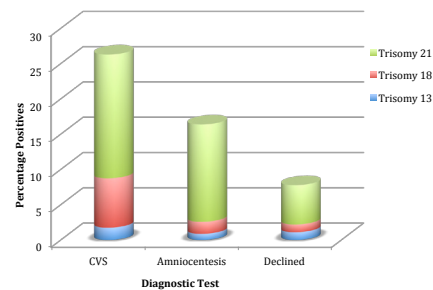
Edwards Syndrome Diagnoses at Ealing Hospital			
Year	All	Prenatal (%)	Live Births Reported
2009-2010	3	3 (100)	0
2010-2011	2	1 (50)	1
2011-2012	2	2 (100)	0
2012-2013	2	0	1
<b>Total</b>	<b>9</b>	<b>5 (55)</b>	<b>2</b>

Patau Syndrome Diagnoses at Ealing Hospital			
Year	All	Prenatal (%)	Live Births Reported
2009-2010	0	0	0
2010-2011	1	0	0
2011-2012	3	3 (100)	1
2012-2013	1	0	0
<b>Total</b>	<b>5</b>	<b>3(60)</b>	<b>1</b>

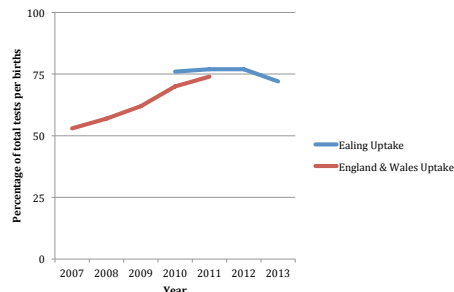
Edwards Syndrome Diagnoses in England and Wales from 2004 to 2012			
Year	All	Prenatal (%)	Live Births Reported
2004	356	320 (90)	38
2005	426	383 (90)	40
2006	451	394 (87)	67
2007	482	441 (91)	54
2008	488	450 (92)	46
2009	520	471 (91)	52
2010	538	484 (90)	65
2011	510	471 (92)	42
2012	526	465 (88)	66
<b>Total</b>	<b>4297</b>	<b>3879 (90)</b>	<b>470</b>

Patau Syndrome Diagnoses in England & Wales from 2004 to 2012			
Year	All	Prenatal (%)	Live Births Reported
2004	147	134 (91)	15
2005	154	135 (88)	24
2006	190	172 (91)	25
2007	213	188 (88)	27
2008	188	170 (90)	24
2009	176	150 (85)	27
2010	221	198 (90)	28
2011	196	172 (88)	23
2012	229	213 (93)	21
<b>Total</b>	<b>1714</b>	<b>1532 (89)</b>	<b>214</b>

## Ratio of Trisomies



## Screening Uptake



Down Syndrome Diagnoses & Outcomes at Ealing Hospital 2009-2013						
Year	All	Prenatal (%)	Live Births Reported	Outcome of Prenatal Diagnoses (%)		
				Termination	Miscarriage/Stillbirth	Live Births
2009-2010	9	7 (78)	3	86	0	14
2010-2011	10	8 (80)	2	100	0	0
2011-2012	7	6 (86)	2	83	0	17
2012-2013	9	5 (56)	4	100	0	0
<b>Total</b>	<b>35</b>	<b>26(74)</b>	<b>11</b>	<b>92.75</b>	<b>0</b>	<b>7.75</b>

Down Syndrome Diagnoses & Outcomes in England and Wales from 2004 to 2012						
Year	All	Prenatal (%)	Live Births Reported	Termination	Miscarriage/Stillbirth	Live Births
2004	1610	978 (61)	660	91	2	7
2005	1751	1040 (59)	740	92	2	6
2006	1836	1109 (60)	758	91	3	6
2007	1789	1106 (62)	716	92	2	6
2008	1844	1128 (61)	750	91	2	7
2009	1927	1193 (62)	787	90	3	8
2010	1911	1219 (64)	728	92	1	6
2011	1901	1232 (65)	733	89	2	9
2012	1982	1259 (64)	765	90	3	7
<b>Total</b>	<b>16551</b>	<b>10264 (62)</b>	<b>6637</b>	<b>91</b>	<b>2</b>	<b>7</b>

## Discussion

Because the national uptake of Down's syndrome in England and Wales is not collected, it was calculated based on total number of tests in the presence of total number of births. The uptake of Downs syndrome screening in England has increased from 53% in 2007, 57% in 2008, 62% in 2009, 60% in 2010, and 74% in 2011. Ealing Hospital uptake stayed relatively consistent with 76% in 2010, 77% in 2011, 77% in 2012, and 72% in 2013. An analysis of offering and uptake from 2008 showed 65% of women report undergoing screening, 89% reported being offered screening, and 69% report taking up the offer of screening. There was no evidence of a difference in offering and uptake for women living in a deprived areas compared to the others. Asian women were less likely to report a screening offer compared to White women and were less likely to undergo screening when offered. A factor that may influence uptake is patients and healthcare providers viewing screening as routine, which eliminates patients understanding they have a choice to accept or decline. Lack of information to parents also impacts decisions about screening. The knowledge, skills, and attitudes of healthcare professionals may not always be suitable to assist informed choices of the patients.

During 2010, there were changes made to the Down's syndrome screening. First trimester combined test gestational age range for nuchal translucency is from 11+2 to 14+1 weeks (previously up to 13+6 weeks), and nuchal translucency should be when crown rump length is between 45 mm and 84 mm (previously up to 80 mm). There was also the enactment of quadruple testing for patients that present after 14 weeks instead of the triple test. The cut off for high risk of screening tests changed from 1:250 to 1:150.

According to the 2012 NDSCR, 64% of Downs syndrome diagnoses were made prenatally. The type of screening used to assess the patient reflects the tissue used for prenatal diagnosis with 58% of women having CVS, 36% having amniocentesis, and 1% unspecified. During the timeframe 2009-2012 at Ealing Hospital, 257 women were offered diagnostic testing. Of these, 57 (22%) had chorionic villus sampling (CVS), 116 (45%) had amniocentesis performed whilst 88 (34%) declined additional diagnostic testing. With the results, 83% of Downs syndrome was diagnosed prenatally.

Between April 1, 2009 and March 31, 2013, eleven thousand nine hundred and ninety nine babies were born at Ealing hospital. There were three cases of Patau syndrome and five of Edwards syndrome with a prenatal diagnosis percentage of 60% and 55%, respectively. According to the NDSCR, there were 477 Edwards syndrome diagnoses, of which 431 (90%) were made prenatally and 190 Patau syndrome diagnoses, of which 170 (90%) were made prenatally. The NDSCR 2012 annual report shows 64% of Downs being diagnosed prenatally and 36% postnatally. At Ealing Hospital, 35 Downs syndrome diagnoses were made, of which 26 (74%) prenatally.

According to the NDSCR 2012 annual report, over 90% of women prenatally diagnosed Downs syndrome underwent a termination of pregnancy. Of the women prenatally diagnosed with Downs syndrome at Ealing Hospital 92% had a termination of pregnancy. There was insignificant data from Ealing Hospital to draw conclusions about the outcome of Edwards and Patau syndromes.

## Conclusion

Although the national uptake of Downs syndrome is increasing, Ealing Hospital's uptake has remained higher and more consistent from 2009 to 2013. This could be a result of enforcement of protocol for informing patients of their rights about screening. Ealing Hospital also had a higher percentage of women diagnosed with Downs prenatally than the national figures. The percentage of termination due to prenatally diagnosed Downs was comparable between Ealing Hospital and England and Wales.

## References

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