

Trends in prevalence of Down’s syndrome in Scotland (2000-2021): a population-based study

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Introduction

Down’s syndrome (DS) is the most common chromosomal condition diagnosed during pregnancy, with a total birth prevalence of **18·4 per 10,000 total births in Scotland**.¹

Screening for DS in pregnancy has been nationally coordinated in Scotland since 2001.² There have been many changes to the screening pathway, most recently with the introduction of **non-invasive prenatal testing (NIPT)** as a secondary screening test in September 2020.³ NIPT is offered as an additional, and more accurate, screening test to women with a pregnancy identified as higher chance for DS by first line screening. To assess the impact of implementing NIPT on screening decisions and pregnancy outcomes, it is important to first understand the baseline trends in total birth and live birth prevalence of DS.

To date, comprehensive and comparable analysis of trends in prevalence of DS and associations with specific socio-demographic factors are unavailable in Scotland. We aimed to understand the long-term trends in **total and live birth prevalence of DS in Scotland (2000-2021)** and explore associations between DS prevalence and socio-demographic factors.

Methods

A **retrospective observational population-based study** of total and live birth prevalence of Down’s syndrome in Scotland.

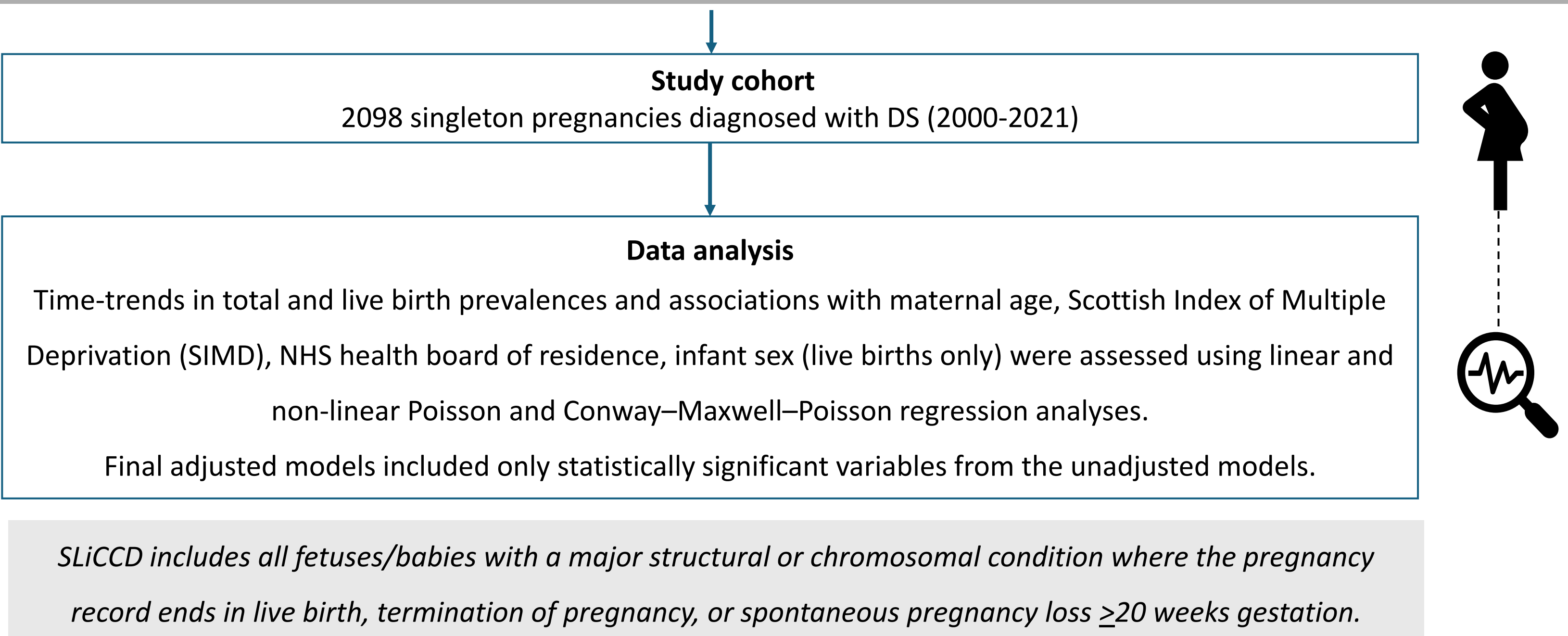
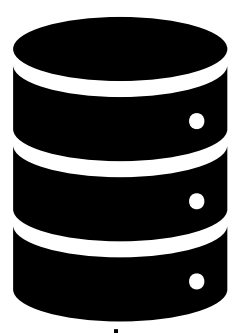
Prevalence rate ratios (PRR), 95% confidence intervals (CI) and p-values reported. All analyses conducted in R (v4.2.1).

Dataset

Scottish Linked Congenital Conditions Dataset (SLiCCD)

Linkage of source datasets: hospital maternity care records; termination of pregnancy notifications; neonatal care discharge records and general hospital discharge records; perinatal death enhanced surveillance records, National Records of Scotland (NRS) statutory live births, stillbirths and infant deaths⁴

Source: Public Health Scotland



SLiCCD includes all fetuses/babies with a major structural or chromosomal condition where the pregnancy record ends in live birth, termination of pregnancy, or spontaneous pregnancy loss ≥ 20 weeks gestation.

Results

2098 singleton pregnancies with DS

ending in Scotland between 2000-2021.

- 1135 live births (54%), 82 stillbirths and late fetal losses (4%), 881 terminations of pregnancy (42%)

Total birth prevalence of DS was 17·43

(16·69, 18·19) per 10,000 total births.

- Evidence of non-linear time trend (Fig.1a): increasing between 2013-2015 [PRR:1·13 (95%CI:1·07,1·19);p<0·001] and decreasing between 2016–2021 [PRR:0·95 (95%CI:0·92,0·98);p=0·002].

Live birth prevalence of DS was 9·48

(8·93, 10·04) per 10,000 live births

- No evidence of a time trend [PRR:1·00(95% CI 0·99, 1·01); p=0·66] (Fig.1b).

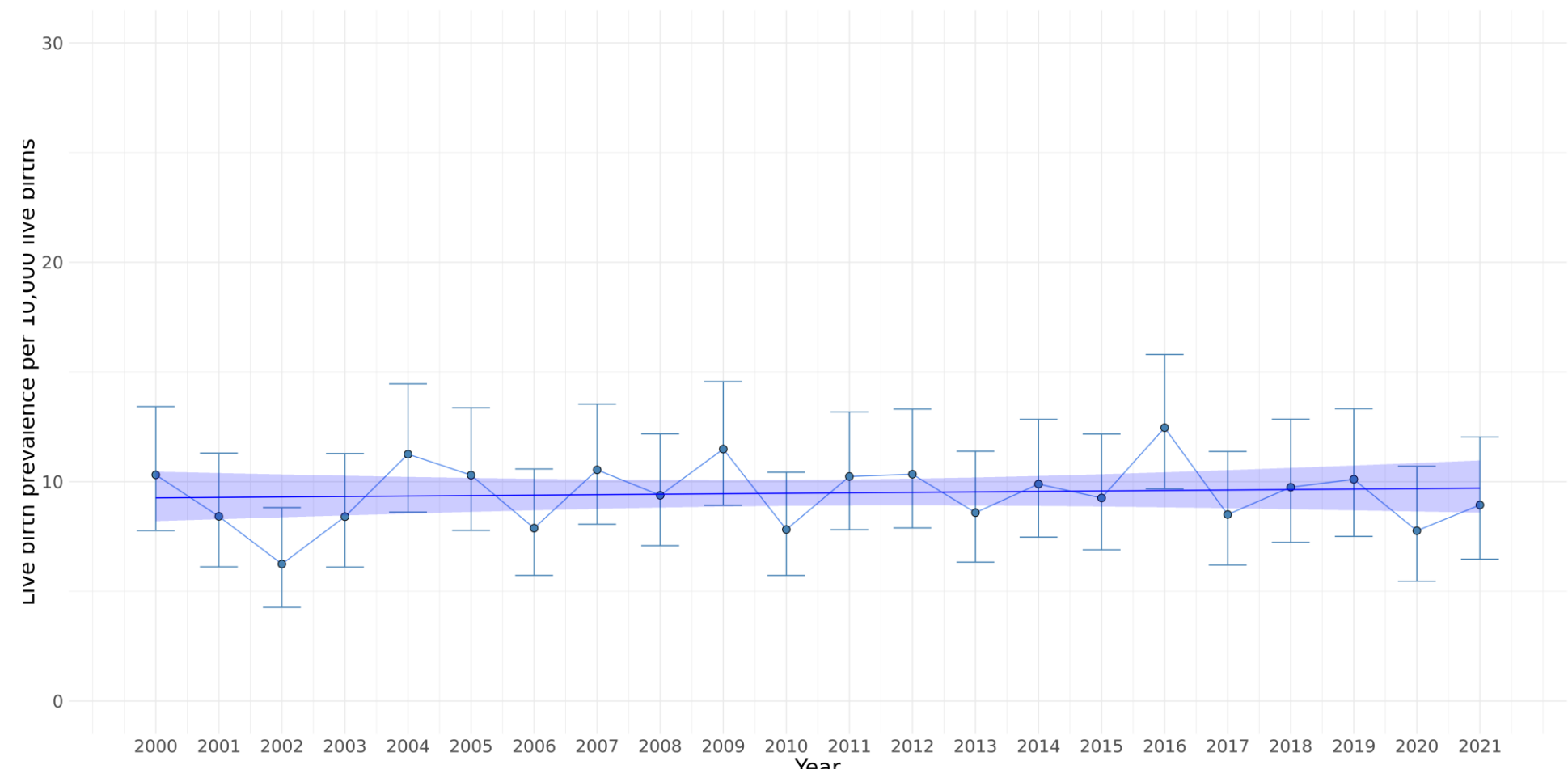
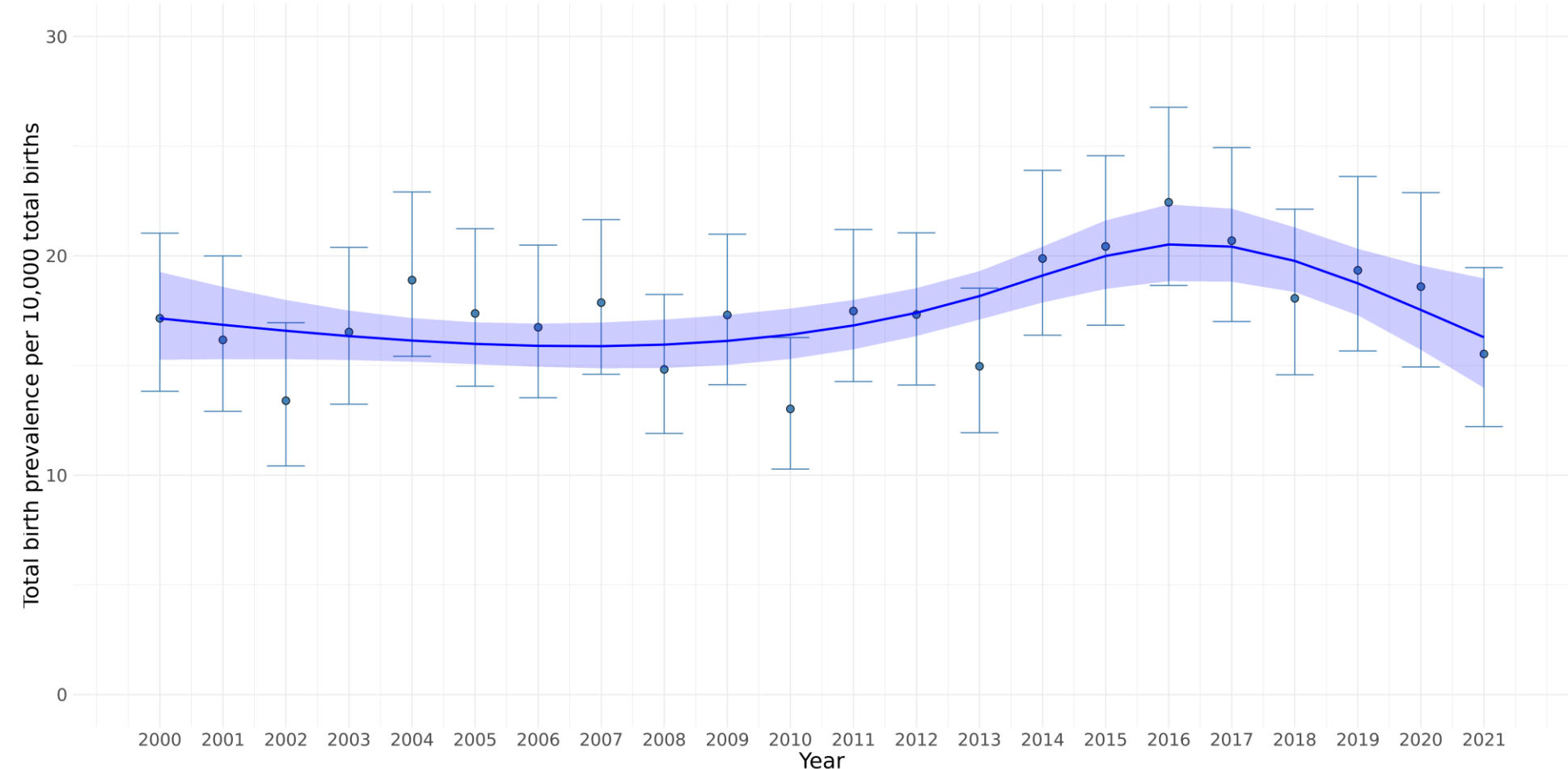


Fig.1: Total and live birth prevalence of Down’s syndrome in Scotland (2000-2021).

(a) Total birth prevalence of DS. Non-linear time trend (4 knots at 2000,2013,2016, 2021). Crude total birth prevalence for each year (black points).

(b) Live birth prevalence of DS. Linear time trend. Crude live birth prevalence for each year and 95% CIs included.

Both figures include model estimated mean total or live birth prevalence (blue line) and 95% CIs (blue ribbon) per year of pregnancy end.

Table 1: Final adjusted COM-Poisson regression model for total and live birth prevalence of Down’s syndrome in Scotland (2000-2021). * P value significance <0.05

	Total birth prevalence of DS		Live birth prevalence of DS	
	Adjusted final PRR (95% CI)	P value	Adjusted final PRR (95% CI)	P value
Maternal age				
Ref - <25	1	-	1	-
25-29	1·07 (0·88, 1·30)	0·51	0·93 (0·76, 1·14)	0·52
30-34	1·93 (1·63, 2·29)	<0·001*	1·45 (1·21, 1·73)	<0·001*
35-39	5·37 (4·56, 6·36)	<0·001*	3·46 (2·92, 4·10)	<0·001*
40+	13·59 (11·34, 16·22)	<0·001*	7·24 (5·93, 8·85)	<0·001*
SIMD				
Ref – 1 (most deprived)	1	-	-	-
2	1·07 (0·92, 1·23)	0·36	-	-
3	0·98 (0·84, 1·14)	0·81	-	-
4	1·00 (0·86, 1·16)	1·00	-	-
5 (least deprived)	1·01 (0·87, 1·17)	0·89	-	-
NHS health board of residence				
Ref - NHS Ayrshire and Arran	1	-	1	-
NHS Borders	0·50 (0·32, 0·80)	<0·001*	0·44 (0·24, 0·77)	0·005*
NHS Dumfries and Galloway	1·14 (0·83, 1·57)	0·42	0·90 (0·61, 1·35)	0·64
NHS Fife	1·13 (0·89, 1·42)	0·33	0·79 (0·59, 1·05)	0·10
NHS Forth Valley	0·94 (0·73, 1·22)	0·66	0·9 (0·68, 1·21)	0·52
NHS Grampian	1·22 (0·98, 1·51)	0·07	0·71 (0·54, 0·92)	0·01*
NHS Greater Glasgow and Clyde	0·75 (0·61, 0·91)	0·01*	0·86 (0·69, 1·08)	0·20
NHS Highland	1·20 (0·93, 1·52)	0·15	1·17 (0·90, 1·54)	0·25
NHS Island Boards	0·83 (0·53, 1·30)	0·40	0·48 (0·25, 0·94)	0·03*
NHS Lanarkshire	0·77 (0·62, 0·96)	0·02*	0·80 (0·63, 1·03)	0·08
NHS Lothian	1·06 (0·87, 1·30)	0·58	0·85 (0·68, 1·08)	0·19
NHS Tayside	1·13 (0·9, 1·42)	0·32	0·97 (0·74, 1·27)	0·81

Conclusion

This study provides detailed and up-to-date population-based insights into the trends and socio-demographic factors associated with DS prevalence in Scotland, with maternal age and NHS health board of residence being significantly associated with total and live birth prevalence of DS in Scotland. Monitoring these trends is important to inform policy and appropriately targeted healthcare planning.

Acknowledgments

ES is funded by Medical Research Scotland PhD studentship (PHD-50200-2020)

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