Trisomy 21 prevalence and socioeconomic status, in Victoria 2015-16

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Introduction

Background:

- Down Syndrome (also known as trisomy 21 or T21) is the most common chromosomal cause of intellectual disability in children
- This study was initiated in the context of low reported rates of T21[1], however a recent government report estimates T21 prevalence rate to be 3.0 per 1000 pregnancies[2]
- It is recommended that prenatal screening be offered to women irrespective of age and background
- Known barriers to being offered or accepting prenatal testing in Australia are younger age, non-English speaking, Indigenous or rural/ geographically isolated

Objectives:

- Provide up-to-date prevalence estimates of the common autosomal trisomies in Victoria
- Explore the association between timing of T21 diagnosis, maternal age and socioeconomic status (measured by IRSAD quintile)

Methods

- All women in Victoria who received a cytogenetic diagnosis of trisomy T21 (T21), trisomy 18 (T18) or trisomy 13 (T13) in 2015-16 were included
- Datasets were obtained from the four cytogenetic laboratories in Victoria
- Prenatal dataset included diagnoses via amniocentesis, chorionic villus sampling (CVS) or following termination of pregnancy (TOP)
- Postnatal dataset included diagnoses in live births and miscarriages/stillbirths
- Screening dataset included combined first trimester screening, second trimester serum screening and non invasive prenatal testing
- Victorian birth numbers were obtained from The Consultative Council on Obstetric and Paediatric Mortality and Morbidity (CCOPMM) reports and the Australian Bureau of Statistics
- Record linkage was performed via LinkageWiz to identify duplicate prenatal and postnatal tests for the same pregnancy - these were assigned to prenatal group only
- Manual linkage was performed to match postnatally diagnosed T21 births with screening
- Socioeconomic status was assigned using Index of Relative Socio-economic Advantage and Disadvantage (IRSAD) from maternal postcode
- Chi^2 testing and logistic regression were performed in STATA v14

Results

- A total of 3662 prenatal and 6250 postnatal diagnostic tests were performed in Victoria during 2015-16
- There were 817 cases of the three common autosomal trisomies (517 prenatal and 300 postnatal diagnoses), giving a combined total prevalence of 5.0 /1000 births (Table 1)
- 17 pregnancies received both a prenatal and a postnatal diagnostic test for the same pregnancy
- Among the T21 group, 12.8% were diagnosed in live infants, 67.4% during pregnancy and 19.8% following miscarriage/ stillbirth
- Among the T18 and T13 cases, only 0.7% and 2.8% of diagnoses were made in live infants
- Of the women that had a T21 diagnosis, there was a significant association between timing of diagnosis and IRSAD quintile (x^2 trend = 15.6, p = <0.01). The proportion of diagnoses in live newborns compared to prenatal diagnoses, decreased with increasing socioeconomic advantage (Fig 1)
- The live birth prevalence of T21 also decreased significantly with increasing socioeconomic advantage (Fig 2)
- There were 73 postnatally diagnosed live T21 births. Of these, 76.7% did not utilise any prenatal screening and only 4.1% received a diagnostic confirmation prenatally
- Women aged 35+ made up 67.7% of women diagnosed with a T21 affected pregnancy

Table 1. Prevalence of T21, T18 and T13 in Victoria, 2015-2016

	T21	T18	T13	Total
Total cases (no.)	577	139	107	823
Total prevalence / 1000 pregnancies[a]	3.6	0.9	0.7	5.1
Live births (no.)	85	2	3	90
Live births / 1000 live births[b]	0.53	0.01	0.02	0.57

Fig 1. Prenatal T21 diagnosis versus newborn T21 diagnosis

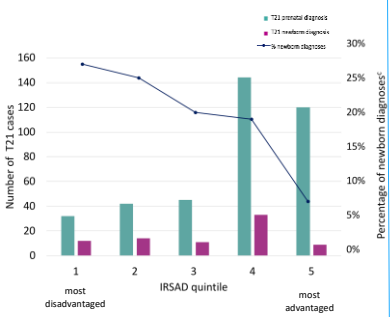
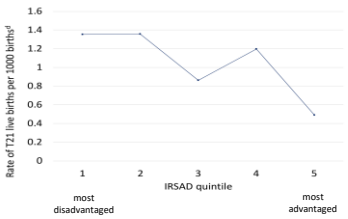


Fig 2. Rate of T21 live births per 1000 live births in Victoria by IRSAD quintile



a. Denominator used to calculate rate per 1000 reported pregnancies (>20 weeks gestation) in Victoria in 2015-16 was 162 192[3] (Table 1)
b. Denominator used to calculate rate per 1000 live births in Victoria in 2015-16 was 158 870[3] (Table 1)
c. No. of newborn diagnosis / (no. of newborn diagnosis + no. of prenatal diagnosis) (Fig 1)
d. No. of T21 live births / all live births, in Victoria, by quintile[4] (Fig 2)

Discussion/Conclusion

- These results highlight the importance of linking prenatal and postnatal cytogenetic datasets to:
  1) accurately report prevalence
  2) compare characteristics of women who receive a prenatal versus a postnatal diagnosis
  3) gain an understanding of pathways through pregnancy
- The prevalence of T21 in Victoria was 3.5 per 1000 pregnancies, which is higher than previously reported[1,2]
- Only 12.8% of all diagnoses of T21 occur in live births
- The majority of live T21 births were not screened prenatally and very few received prenatal diagnostic confirmation
- The significant association between socioeconomic disadvantage and having a live born infant with T21, raises important questions about access to, and utilization of, prenatal testing services
- This association may have multiple causes, including patient factors (such as personal preference or attitudes toward prenatal testing), practitioner factors (such as lack of an offer of prenatal screening/diagnostic test) or external factors such as financial or geographical barriers to services

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