the partumpost

VCGS Leaders in Genetic Health

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Prenatal diagnostic trends

This issue of *partumpost* explores the long term trends of prenatal diagnosis in a summary of the Report on Prenatal Diagnostic Testing in Victoria in 2017 produced by the Reproductive Epidemiology group from the Murdoch Children's Research Institute.

Invasive procedures are declining in Victoria

Prenatal diagnostic testing started in the late 1970s. The more recent decline in the number of invasive procedures is mostly in response to non-invasive or cell-free DNA screening. *See figure 1.* Non-invasive prenatal screening is a more accurate screening test and therefore, less women are referred for invasive procedures and diagnostic testing.

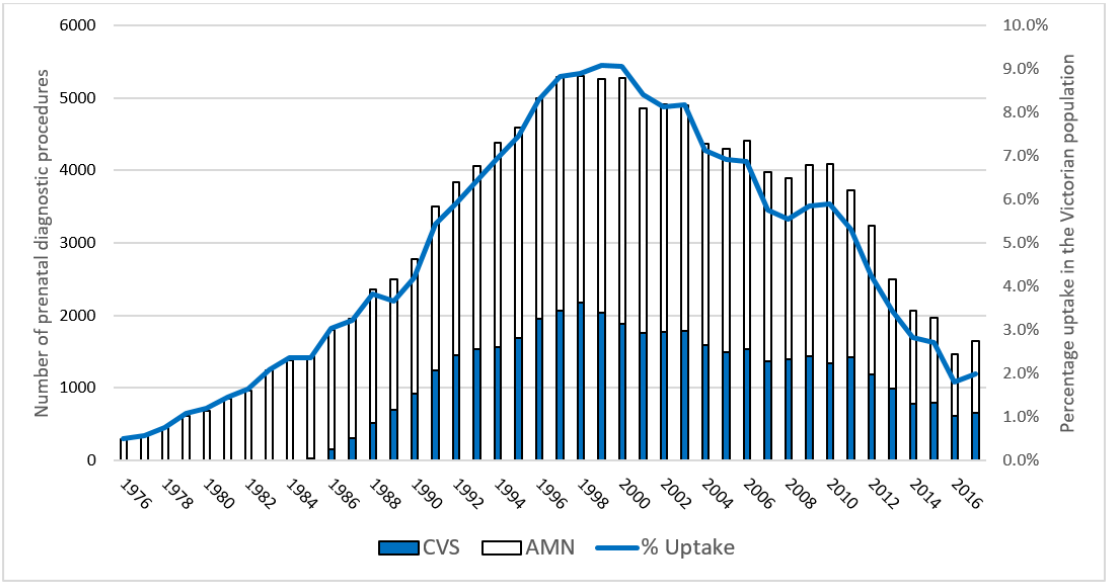

Figure 1. Prenatal diagnostic procedures

The most common indication for diagnostic testing is an abnormal ultrasound. This is followed by high risk screening results from either combined first trimester screening or cell-free DNA screening (NIPT). *See figure 2.*

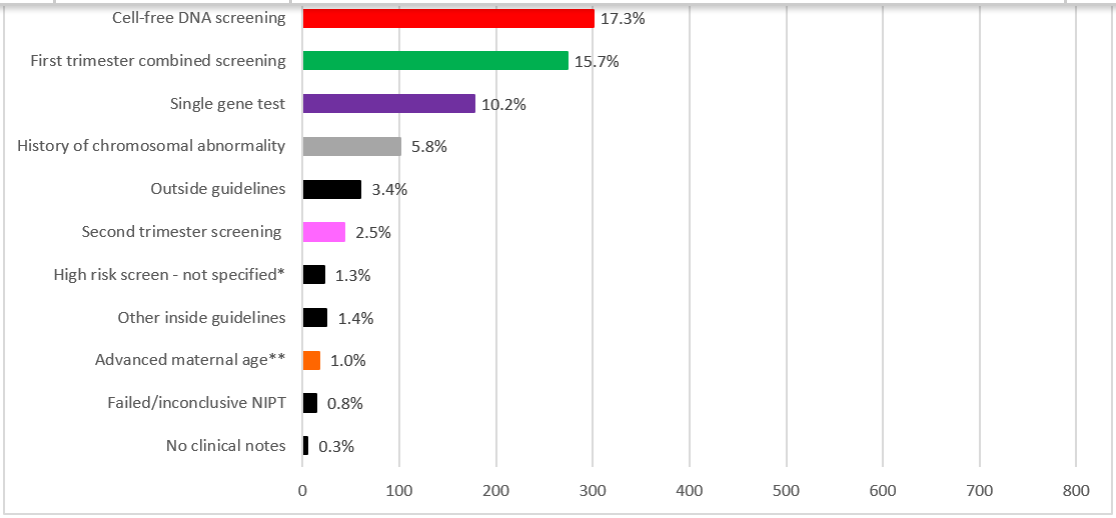

Figure 2. Indications for prenatal diagnosis

Chromosome microarray is now used for >85% of prenatal diagnosis in Victoria, regardless of indication. *See figure 3.*

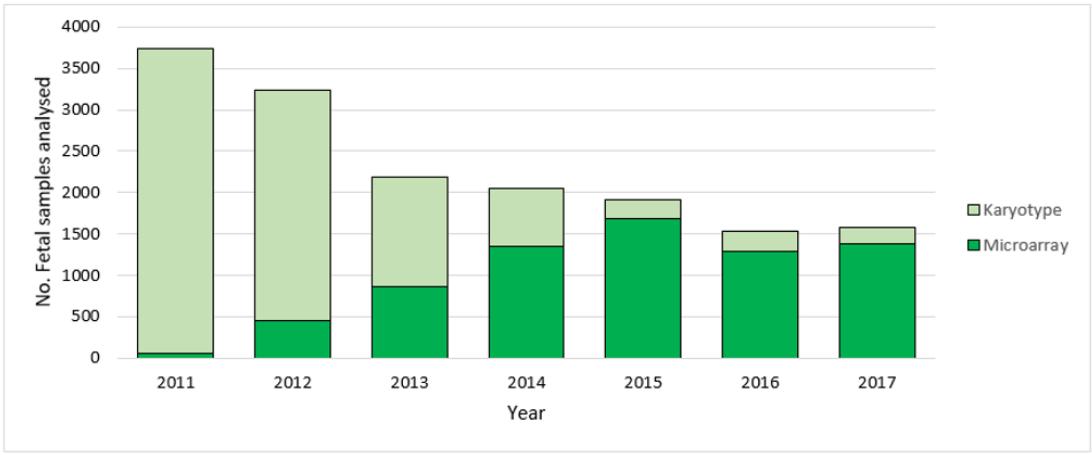

Figure 3. Prenatal sample types

The prenatal prevalence of trisomy 21 as a proportion of all births in 2017 (n=80,934) was 1 in 426. The prevalence of trisomy 18, trisomy 13 and total sex chromosome abnormalities were 1 in 1471, 4496 and 1527 respectively. *See figure 4.*

Karyotype/microarray result	2014 n=2046	2015 n=1957	2016 n=1468	2017 N=1643
Normal/benign variant	1548	1427	1037	1152
Major chromosome abnormalities	**369**	**394**	**363**	**394**
Trisomy 21	176	204	183	190
Trisomy 18	49	42	44	55
Trisomy 13	21	15	25	18
Other autosomal aneuploidy, polyploidy	22	21	9	14
Pathogenic copy number variation	39	39	29	44
Other abnormalities	62	73	73	73
Variant of unknown/uncertain significance	108	126	68	93

Figure 4. Prenatal diagnostic results

See figure 5.

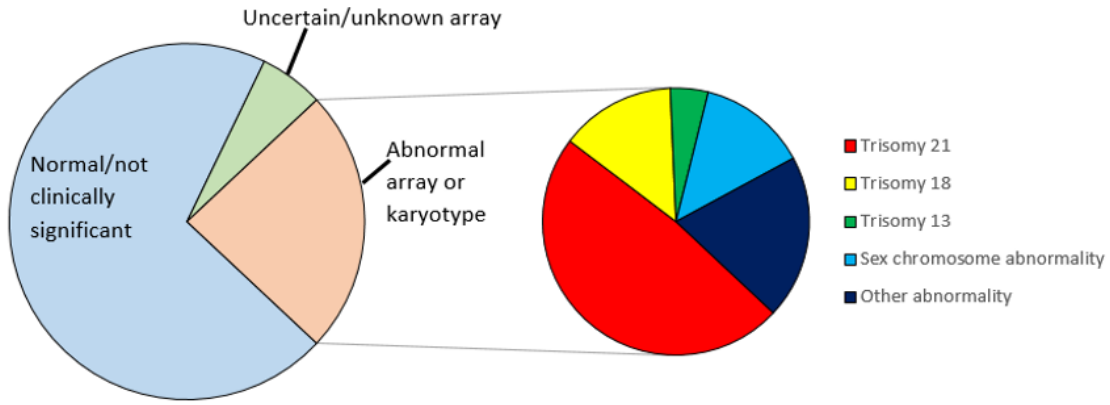

Figure 5. Outcomes of prenatal diagnostic tests

POULTON, A., HUI, L. AND HALLIDAY, J. (2019). REPORT ON PRENATAL DIAGNOSTIC TESTING IN VICTORIA IN 2017. MURDOCH CHILDREN'S RESEARCH INSTITUTE

Read the full Report on Prenatal Diagnostic Testing in Victoria in 2017 or for more information please **contact us**.

Phone: 1300 118 247 or email: vcgs@vcgs.org.au

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