

PERSONALISED ANTENATAL CARE OF PREGNANCIES SUSPECTED OR DIAGNOSED WITH DOWN SYNDROME

This document has been written for the use in the fetal medicine department at St George's University Hospitals NHS Foundation Trust. This document offers evidence-based guidance to obstetricians and midwives about the on-going management of pregnancies suspected or diagnosed with Down syndrome. There is currently no national antenatal guideline for the on-going care, and therefore guidance and care may vary depending on trust or region.

AIM

Women who have had a high chance non-invasive prenatal trisomy screening result or those that have undergone an invasive test to confirm Down syndrome require personalised antenatal care, to optimise maternal and neonatal outcomes. Women should be provided with information and support throughout the antenatal period, as well as appropriate referral for medical and social support. These recommendations reflect the increased risk of associated structural fetal problems and obstetrical risks. This resource outlines the additional midwifery, medical and psychological support that is recommended to deliver personalised medical care.

ANTENATAL CARE

All pregnant women whose babies are known to have or suspected to have Down syndrome should be offered the opportunity to be case-loaded by a named midwife or team. The antenatal care pathway should be discussed and personalised to the needs of the parents, the care should, at a minimum, follow the nulliparous pathway of a minimum of 10 visits within the pregnancy regardless of parity, if accepted by women. Wherever possible, appointments should be incorporated with routine tests and investigations to minimise inconvenience. Women with additional maternal or fetal risk factors should be guided as per national and local recommendations for consultant and specialist care pathways (e.g. Diabetes, BMI and Hypertension).

ASSESSMENT FOR FETAL STRUCTURAL PROBLEMS

Concern:

Newborns with Down syndrome have a significantly increased risk of structural problems. About 50% of babies with Down syndrome will have a cardiac defect, which may sometimes be difficult to diagnose prenatally. Other anomalies that are more common with Down syndrome include duodenal atresia, duodenal stenosis, renal abnormalities and Hirschsprung's disease.

Improving outcome:

Prenatal detection of any associated structural problems will help to inform parents and prepare carers about any anticipated issues in the newborn period.

Recommendations:

- Detailed ultrasound and fetal echocardiogram at 18 to 22 weeks.
- Repeat detailed ultrasound at 28 to 32 weeks to recheck cardiac views and assess for evidence of upper gastrointestinal obstruction, hydrothorax and fetal hydrops.

ASSESSMENT FOR THE RISK OF PREMATUREITY

Concern:

Pregnancies where the fetus has or suspected of having Down syndrome face a substantial chance of early delivery, with about 20% delivering prior to 37 weeks. Even when medically indicated births (such as for preeclampsia) have been excluded, the residual risk for spontaneous preterm birth remains higher than the background population risk of preterm birth – usually secondary to polyhydramnios from fetal duodenal atresia.

Improving outcome:

Prenatal detection of polyhydramnios related to duodenal atresia is amenable to prenatal therapy (amniodrainage) if the pregnancy is considered to be at increased risk of preterm birth.

Recommendations:

- Ultrasound assessment for women presenting with symptoms of contractions or abdominal distention after 28 weeks' gestation to exclude the diagnosis of upper gastrointestinal obstruction and associated polyhydramnios.

ASSESSMENT FOR THE RISK OF PLACENTAL DYSFUNCTION

Concern:

Pregnancies where the fetus has or suspected of having with Down syndrome face an increased risk of placental dysfunction and the consequences of fetal growth restriction and stillbirth. Even in the absence of placental dysfunction, there is an excess risk of stillbirth with expectant management beyond 39 weeks' gestation.

Improving outcome:

Prenatal detection of placental dysfunction will allow monitoring and timely scheduled birth in order to avoid the increased risk of stillbirth.

Recommendations:

- Ultrasound scans at 28-32 and 36-37 weeks' gestation to assess fetal growth and Doppler blood flows – with appropriate subsequent monitoring and management if there are concerns about placental dysfunction.
- As with all pregnant women, parents should be advised to self-refer immediately to their maternity unit if they have concerns with reduction in fetal movements.
- A consultation with an Obstetrician is recommended to discuss the option of elective induction of labour from 39 weeks' gestation as well as place of birth.

EMOTIONAL AND PSYCHOLOGICAL SUPPORT

Concern:

Parents who receive the diagnosis (or a high chance NIPT result) that their fetus is affected by Down syndrome can struggle to bond with their newborn and can experience difficulty coping and psychological distress. These effects are worsened by negative comments or encounters from healthcare staff at various points in the care pathway.

Improving outcome:

Parents would benefit from use of unbiased language and attitudes and early access to psychological support (both formal and informal). Mindful and considerate use of language when delivering diagnosis and pregnancy care planning could improve outcomes for mother and baby.

Recommendations:

- Early access/sign posting to pre and postnatal psychological support and peer support via organisations listed.

POSTNATAL CONSIDERATIONS

Concern:

Delays in referrals to specialist services can lead to a variety of issues which can affect outcomes for growth, development and wellbeing.

Improving outcome:

Ideally, parents should have the opportunity to meet members of the neonatal team before the birth to discuss the postnatal plan of care. Women who plan to breastfeed may need support with possible issues e.g. sleepiness, poor tone and coordination.

Recommendations:

- Parents value an antenatal appointment with the neonatal team to discuss postnatal care and considerations.
- Women who are planning to breastfeed their babies should be referred to the local breastfeeding support team or lactation consultant. A breastfeeding guide developed by DS-UK should be shared with the parents if appropriate: <https://www.downsyndromeuk.co.uk/docs/Health/12294%20DSUK%20A%20Guide%20To%20Breastfeeding-PRINT.pdf>

REFERENCES

1. Muglu J et al. PLoS Med. 2019;16: e1002838.
2. Rasmussen SA et al. J Pediatr. 2006;148:806-812.
3. Rutter, T. et al. Journal of medical genetics part A, 2025: e64206
4. Rutter, T. et al. *Journal of Applied Research in Intellectual Disabilities* 2025: e70160
5. Skotko BG et al. Am J Med Genet A. 2009; 149:2361-7.
6. Sparks TN et al. Prenatal Diagn. 2016; 36:368-74.
7. Van Riper M and Choi H. Genetics in Medicine 2011; 13: 714 – 716.

AUTHORS

Joanne Hargrave BSc RM – NIPT Senior Midwife Coordinator

Basky Thilaganathan MD PhD FRCOG - Professor and Director, Fetal Medicine Unit

ACKNOWLEDGEMENTS

We are grateful for the significant contribution, collaboration and support from Positive about Down syndrome, Down Syndrome UK, the Down Syndrome Research Foundation UK, and Angie Bowles (Independent Midwife) without which we would not have been able to develop this guideline.

