



Perinatal Services BC
An agency of the Provincial Health Services Authority



Management of Pregnancies Diagnosed with Down Syndrome

**Practice Resource for
Health Care Providers**

September 2016

Practice Resource Guide: MANAGEMENT OF PREGNANCIES DIAGNOSED WITH DOWN SYNDROME

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SUMMARY

Prenatal screening and ultrasound allows for most pregnancies with Down syndrome to be identified prenatally. Care of the family who have received a diagnosis of Down syndrome should be provided by an interdisciplinary team including clinical geneticists and genetic counsellors, pediatricians, obstetricians, the family's primary care provider and nurses.¹ Initial meetings with the family should be sensitive realizing that the family's dreams for their child have been altered.² Counselling of these pregnant women and their partner should provide accurate, current and balanced information that presents the obstetrical risks and medical concerns associated with Down syndrome as well as the benefits and challenges of raising a child with Down syndrome. Approximately fifty percent of babies with Down syndrome will have a structural abnormality with cardiac defects being the most common. In addition, pregnancies with Down syndrome are at increased risk of prematurity, intrauterine growth restriction, intrauterine fetal demise and neonatal death. Each family's reaction to the diagnosis will be different and one approach will not work for all.³ Informed decision-making is paramount for these families throughout the initial diagnosis, obstetrical management, and birth of their child.

This practice resource has been developed to optimize maternal and newborn outcomes in this patient population.

Recommendations for the comprehensive evaluation and management of pregnancies diagnosed with Down syndrome include:

- Detailed ultrasound and fetal echocardiogram at 18–20 weeks gestation, or at time of diagnosis of Down syndrome if later;
- Repeat detailed ultrasound at 28–30 weeks to assess for evidence of upper gastrointestinal obstruction, chylothorax, fetal hydrops and IUGR;
- Cervical assessment by endovaginal (EV) scan should be done at the time of the 18–20 weeks ultrasound and at every follow-up ultrasound;
- Consultation with an Obstetrician or Maternal Fetal Medicine specialist if cervical length less than 2.5 cm;
- Ultrasound done at 28–30 weeks to screen for fetal anomalies should pay special attention to fetal biometry and the interval growth since the last scan;
- Ultrasound at 36 weeks to reassess for IUGR;

- Consultation with an Obstetrician or Maternal Fetal Medicine specialist is recommended as well as increased fetal surveillance if intrauterine growth restriction is diagnosed;
- Timing of delivery should be based on results of fetal assessment if IUGR present;
- Induction of labour is recommended for pregnancies reaching 40 weeks gestation;
- Pregnancies with no identified fetal abnormality should be delivered in a hospital providing at a minimum Tier 2 level of care. Pregnancies with a prenatally identified fetal structural abnormality should be delivered at BC Women's Hospital (Tier 4);
- Intrapartum care should be based on maternal and fetal risk factors;
- All newborns with Down syndrome should be assessed by a pediatrician before discharge;
- All newborns with Down syndrome should have an echocardiogram done within the first few months of life (or sooner if clinically indicated) even if a prenatal fetal echocardiogram was normal;
- Breastfeeding and skin-to-skin should be actively supported;
- Referral for a public health visit should be made at discharge.

INTRODUCTION

Prenatal screening by Serum Integrated Prenatal Screen (SIPS)/Integrated Prenatal Screen (IPS)/Quad, Non Invasive Prenatal Testing (NIPT) and ultrasound allows for the identification of most pregnancies with Down syndrome. Counselling of these pregnant women and their partner should provide accurate, current and balanced information that presents the obstetrical risks (increased risk of prematurity, intrauterine growth restriction {IUGR} and intrauterine fetal demise {IUFD}) and medical concerns associated with Down syndrome (including increased rate of neonatal death related to prematurity and major structural abnormality) as well as the benefits and challenges of raising a child with Down syndrome. Couples should be informed of their option to continue or terminate the pregnancy. They should also be given the option of a consultation in medical genetics. Of particular significance is the delivery of the diagnosis and ongoing information to the couples. Clinical Practice Guidelines⁴ describe many topics that care providers must discuss with parents after the diagnosis of Down syndrome but few remind providers to display compassion as the parents may be grieving the loss of a baby they thought they were having and trying to process what lies ahead.⁵ Acknowledging that the family now has a change of plan rather than approaching it as a tragedy can greatly impact those initial discussions². The initial contact with a care provider will leave lasting memories for the family.

Comprehensive evaluation and management of ongoing pregnancies should reflect the increased risk of congenital malformations and obstetrical risks associated with the diagnosis of Down syndrome in order to optimize maternal and newborn outcomes. This resource was developed after a review of the obstetrical and newborn outcomes of a cohort of 295 pregnancies that continued past 20 weeks gestation and had prenatal or postnatal diagnosis of Down syndrome made between the calendar years 2007 and 2013. The obstetrical and neonatal data was extracted from the BC Perinatal Data Registry⁶ after ethics approval from the University of British Columbia Research Ethics Board. A review of the literature was also carried out.

ASSESSMENT FOR THE PRESENCE OF FETAL STRUCTURAL ABNORMALITIES

Newborns with Down syndrome have a significantly increased risk of structural abnormalities. Approximately 50 percent will have a cardiac defect and not all of these defects will be detected prenatally. Other anomalies more common with Down syndrome include duodenal atresia, duodenal stenosis, renal abnormalities and Hirschsprung's disease.

Recommendations

- Detailed ultrasound and fetal echocardiogram at 18–20 weeks gestation or at time of diagnosis of Down syndrome if later.
- Repeat detailed ultrasound at 28–30 weeks to assess for evidence of upper gastrointestinal obstruction, chylothorax, fetal hydrops and IUGR.

ASSESSMENT FOR THE RISK OF PREMATUREITY

Amongst the cohort of 295 pregnancies, two pregnancies resulted in intrauterine fetal demise prior to labour, one at 26 weeks and one at 28 weeks (0.7 percent). Amongst the remaining 293 pregnancies, 7 percent resulted in a premature birth less than 34 weeks gestation (compared to 2.4 percent in the 2012/2013 BC population, BC Perinatal Data Registry report) and 22.5 percent resulted in a premature birth between 34–36⁺⁶ weeks (compared to 7.6 percent in the 2012/2013 BC population, BC Perinatal Data Registry report). If one excludes the pregnancies with IUGR, maternal hypertension, pre-eclampsia or HELLP which may have been in part responsible for the premature delivery, there remains a 4.4 percent risk of premature delivery before 34 weeks and 17.4 percent risk of premature delivery between 34–36⁺⁶ weeks. These frequencies are approximately twice the population frequencies which would indicate an increased risk of prematurity in otherwise uncomplicated Down syndrome pregnancies.

Recommendations

- Cervical assessment by endovaginal (EV) scan should be done at the time of the 18–20 weeks ultrasound and at every follow-up ultrasound.
- Consultation with an Obstetrician or Maternal Fetal Medicine (MFM) specialist if cervix length less than 2.5 cm.

ASSESSMENT FOR THE RISK OF IUGR

When using standard growth curves, amongst the entire cohort of 293 live births, 13 percent of newborns were Small Gestational Age (SGA) defined as less than the 5th percentile and 7 percent were less than the 3rd percentile. The incidence of SGA and the percentage of SGA newborns diagnosed prenatally as IUGR were studied in different gestational age groups. The results are presented in the table below. Our results indicate

that the incidence of IUGR is higher in newborns born after 41 weeks. Furthermore, the prenatal diagnosis of IUGR is made in a high proportion of SGA newborns born before 37 weeks but in a very small percentage of SGA newborns born after 37 weeks. For example, for newborns born less than 34 weeks gestation, 13 percent were SGA and all were detected prenatally. However, 28 percent of newborns born after 41 weeks were SGA and none were diagnosed prenatally with IUGR.

Incidence of SGA and % of SGA babies detected prenatally as IUGR

| GA | # | IUGR not SGA | IUGR SGA | # SGA <5th by standard growth curves (%) | % of SGA detected by U/S | # SGA <5th by T21 VON growth curves (%) | % of SGA detected by U/S |
|---------------------|-----|--------------|----------|------------------------------------------|--------------------------|-----------------------------------------|--------------------------|
| <34 | 23 | 5 | 3 | 3 (13%) | 100% | 1 (4%) | 100% |
| 34–36 ⁺⁶ | 66 | 6 | 5 | 7 (11%) | 71% | 2 (3%) | 100% |
| 37–40 ⁺⁶ | 199 | 1 | 4 | 26 (13%) | 15% | 9 (4.5%) | 11% |
| 41+ | 7 | 0 | 0 | 2 (28%) | 0% | 2 (28%) | 0% |

When using published anthropometric charts for infants with trisomy 21 developed by the Vermont Oxford Network (VON),⁷ a non-profit voluntary collaboration of health care professionals from US and International neonatal intensive care units, the overall incidence of SGA is as expected (4.7 percent). However, the percent of SGA newborns amongst those delivered at or after 41 weeks is high (28 percent) which may reflect placental insufficiency. Again, the majority of SGA newborns, as per the VON growth charts that were born after 37 weeks, were not diagnosed prenatally as IUGR.

When IUGR is diagnosed, increased fetal surveillance is important with ultrasound monitoring of fetal growth every two weeks. If IUGR is:

- Mild (AC 5 – 9th percentile) – weekly NST, AFI and UA Doppler
- Moderate (AC 1 – 4th percentile) – two × week NST, AFI and UA Doppler
- Severe (AC less than 1st percentile) – three × week NST, AFI and UA Doppler

Definitions: AC – abdominal circumference; NST – non-stress test; AFI – amniotic fluid index; UA – umbilical artery.

Recommendations

- Ultrasound done at 28–30 weeks to screen for fetal anomalies should pay special attention to fetal biometry and the interval growth since the last scan.
- Ultrasound at 36 weeks to reassess for IUGR.
- Consultation with an Obstetrician or Maternal Fetal Medicine specialist is recommended as well as increased fetal surveillance if intrauterine growth restriction is diagnosed.
- Timing of delivery should be based on results of fetal assessment if IUGR present.

PLANS FOR DELIVERY

To reach recommendations regarding timing and appropriate location for delivery of pregnancies with an antenatal diagnosis of Down syndrome, we analyzed the rate of stillbirth, mode of delivery, and level of care required for the 245 newborns with Down syndrome born at 36 weeks or later. Rate of in-hospital neonatal death was also assessed.

Amongst our cohort of 295 pregnancies, there were two stillbirths, one at 26 weeks and one at 28 weeks gestation. This stillbirth rate is not increased compared to the 2012–2013 BC population rate of 9.4 per 1,000 births (BC Perinatal Data Registry Report). However, the literature indicates that pregnancies with Down syndrome have a significant increased risk of IUFD. Won et al., reported on a rate of IUFD of 10.2 percent in a cohort of 392 pregnancies prenatally diagnosed with Down syndrome that chose to continue the pregnancy.⁸ Some of those IUFD were between 20–24 weeks gestation and may have represented termination of pregnancies. The rate of IUFD at or after 24 weeks was 5.8 percent and was uniformly distributed throughout gestation after 24 weeks.

The finding in our cohort of a significant increased rate of IUGR in post term pregnancies (28 percent compared to 13 percent) may reflect placental insufficiency which would put these pregnancies at increased risk for stillbirth. Furthermore, excluding the two pregnancies with IUFD, 37 percent of pregnancies were delivered by C/S which is comparable to the rate in the 2012–2013 pregnant population of 31.2 percent (BC Perinatal Data Registry Report). However, in 30 percent of cases, the C/S was done for atypical/abnormal fetal heart rate compared to only 17.4 percent of C/S being performed for that indication in the 2012–2013 pregnant population (BC Perinatal Data Registry Report).

Based on the literature findings of increased stillbirth and our findings of IUGR suggestive of placental insufficiency, induction of labour is recommended for pregnancies reaching 40 weeks gestation. Intrapartum care should be based on maternal and fetal risk factors. If the fetus is IUGR, then protocols for surveillance of IUGR should be followed. The same is true for any other risk factor.

Amongst newborns with Down syndrome and no congenital anomalies born at 36 weeks gestation or later, 14 percent required level 2 care and 4 percent required level 3 care, as defined in 2007–2013. In contrast, 39 percent of newborns with Down syndrome born with anomalies required level 2 and 23 percent required level 3 despite being born at or later than 36 weeks gestation. It is worth noting that the percentage of newborns requiring level 3 care and the number of days spent in level 3 care may be an underestimate as data quality issues related to neonatal intensive care (NICU) days beginning April 2010 to the end of our study period may have led to an under-reporting of stays in NICU.

Amongst the cohort of 293 liveborns, a neonatal death occurred in 7 cases (2.4 percent). These newborns were either born prematurely or had congenital anomalies or both. Amongst the cohort of newborns born before 32 weeks gestation, the rate of neonatal death was 22 percent.

Recommendations

- Induction of labour is recommended for pregnancies reaching 40 weeks gestation.
- Pregnancies with no identified fetal abnormality should be delivered in a hospital providing at a minimum Tier 2 level of care. Pregnancies with a prenatally identified fetal structural abnormality should be delivered at BC Women's Hospital (Tier 4).
- Intrapartum care should be based on maternal and fetal risk factors.

NEWBORN CARE

All newborns with Down syndrome should be assessed by a pediatrician before discharge. Ongoing health supervision information for physicians is outlined in the *Clinical Report – Health Supervision for Children with Down Syndrome*.¹ The nurse will be responsible for ongoing assessments while the family is in the hospital. Some of the characteristics to be aware of when caring for a baby with Down syndrome are respiratory distress or colour changes secondary to cardio-respiratory problems, ruddy colour from polycythemia or marbling of the skin (cutis marmorata), and hypotonia.³

Breastfeeding has additional benefits for a baby with Down syndrome. Along with the extra protection that the antibodies found in human milk offer, breastfeeding is also associated with a decreased risk for obesity.⁹ The sucking action can help develop mouth and tongue coordination and strengthen the jaw and facial muscles. Often, a baby with Down syndrome has feeding difficulties related to sleepiness and poor tone; it is even more evident if there are congenital heart problems. As a result, the baby may need more support of the head, neck and chin during feeding. A lactation consultant can help with feeding difficulties. If poor feeding leads to growth delay, supplementation may be required.² An excellent resource for parents can be found on the Canadian Down Syndrome Society website.¹⁰

Every effort should be made for enabling close proximity as well as ability to touch and make eye contact. Skin-to-skin should be encouraged. Families should be informed that positive interactions like smiling, vocalizations and imitating behaviours will help with their baby's development.²

Discharge planning should include a referral for a public health visit to ensure feeding is going well, in addition to help coordinate any community resources or required follow-up. Public Health nurses provide a continuum of care and should be part of the team caring for the family. A referral to the Infant Development Program of BC is also indicated. Contacts to Down syndrome societies are helpful for parents so they can access these resources when they are ready. A list is provided at the end of this Practice Resource.

Recommendations

- All newborns with Down syndrome should be assessed by a Pediatrician before discharge.
- All newborns with Down syndrome should have an echocardiogram done within the first few months of life (or sooner if clinically indicated) even if a prenatal fetal echocardiogram was normal.
- Breastfeeding and skin-to-skin should be actively supported.
- Referral for a public health visit should be made at discharge.

EMOTIONAL SUPPORT

The diagnosis of Down syndrome for a family may alter the dreams and expectations they held for their child. Although each family will be different in their reaction, encouraging and supporting parents to focus on adapting their expectation can help them when they are confronted with the diagnosis.² A survey of 1,250 women with infants who had Down syndrome found the majority had negative encounters with health care providers at the time of the diagnosis or birth of their child.¹¹ Families require support, education and guidance with the long-term management of their child's health needs, whatever they may be.^{2,5} The care of the family requires a multidisciplinary team approach. The multidisciplinary team may consist of primary care physicians, midwives, pediatrics, pediatric cardiology, nursing (acute and community), occupational therapy, physical therapy, speech therapy, lactation consultants, pediatric ophthalmology and support groups for parents.¹² There will be many factors that are out of the health care providers' control but one thing we can control is our approach. Parents are most helped by an unbiased attitude from health care providers and positive comments about the baby, coupled with emotional support, sensitivity, and hope.¹³

The nurse will often be the primary care provider for the family for labour and during their initial adjustment after the baby is born. At this time, they have the power to affect and change the experience for families. Along with the physical assessments that are required, taking time to congratulate the parents and to talk positively about their newborn as well as calling the baby by name can help their adjustment.²

RESOURCES AND SUPPORT

For Families

Lower Mainland Down Syndrome Society (BC), phone 604-591-2722; website www.lmdss.com

Canadian Down Syndrome Society, phone 1-800-883-5608; email info@cdss.ca; website www.cdss.ca

Down Syndrome Research Foundation (Canada), phone 604-444-3773 or toll-free in Canada 1-888-464-DSRF; website www.dsrf.org

ChildHealth BC; website www.childhealthbc.ca

<http://understandingdownsyndrome.org> from The National Center for Prenatal and Postnatal Resources, Human Development Institute, University of Kentucky

Infant Development Program of BC; website www.idpofbc.ca

For Providers

Genetics Home Reference (US National Library of Medicine); website www.ghr.nlm.nih.gov

Society of Obstetricians and Gynaecologists, Clinical Practice Guidelines (Canada); website www.sogc.ca

BC Prenatal Genetic Screening Program; website www.bcprenatalscreening.ca

Perinatal Services BC; website www.perinatalservicesbc.ca for full electronic copy of this Practice Resource and all other PSBC publications.

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