



Information for Physicians

The following checklist will help you plan the health screenings and resources that are recommended for babies with Down syndrome.

GENETICS

- For all new babies with Down syndrome, please consult the Genetics and Metabolism Fellow on call in the Partners Paging Directory (www.ppd.partners.org).
 - **If the baby is in the NICU**, please consult the genetics fellow as soon as your team feels it is clinically appropriate. We are happy to be involved in the patient's care or provide additional support to the family.
 - **If the baby has a postnatal diagnosis in the newborn nursery**, please consult us as soon as you suspect the diagnosis.
 - **If the baby is in the newborn nursery and already has a prenatal diagnosis**, please consult us anytime before discharge.
- For postnatal diagnosis: Please send FISH and karyotype.
 - FISH and karyotype are completed at the Center for Advanced Molecular Diagnostics, Cytogenetics Laboratory at Brigham and Women's Hospital (BWH).
 - Although the form says "prenatal," it is the correct form to use.
 - Please send 2 – 4 mL (sodium heparin tube) with the completed requisition included in this packet. Please send the blood and requisition to MGH pathology who will further process the sample and see that it gets to the cytogenetics lab at BWH.

DOWN SYNDROME HEALTH SCREENING

- Thyroid function tests
 - The Newborn Screening Program routinely runs T4 on all babies. Babies with Down syndrome may have slightly elevated TSH; therefore, please note the following on the NBS paperwork: "Suspected known Down syndrome diagnosis. Please check TSH and T4."
- Check a CBC with differential
 - This rules out Transient Myeloproliferative Disorder
- Cardiac evaluation:
 - Please consult Cardiology for all babies with Down syndrome within 24 hours of birth. Cardiology will arrange for an echocardiogram, which will be done regardless of whether there is an audible murmur.
 - Prior to Cardiology arriving for consultation, please check an initial ECG, 4-extremity blood pressures, and pre- and post-ductal saturations.
- Car seat evaluation
- Check red reflex

REFERRALS

MassGeneral Hospital Down Syndrome Program

- As part of this consult, we will arrange for a follow-up visit for your patient.
- If you or your patient has any further questions, please call 617-643-8912 or e-mail downsyndrome@partners.org.
- For more information on our program, visit www.massgeneral.org/downsyndrome.

Lactation Support

- Many babies with Down syndrome can successfully breast feed with additional lactation support.
- We would greatly appreciate if you could arrange for a lactation consultant to visit with the mom and baby before discharge.

Massachusetts Down Syndrome Congress (MDSC)

- The MDSC will provide free books and information about Down syndrome to the patient and family.
- The MDSC can also connect the family, if they are interested, with another family who already has a child with Down syndrome and was in similar circumstances. Enclosed in this packet is a consent form that can be used for you to gather the family's permission to share their contact information with the MDSC.
- For more information, visit www.mdsc.org.

Early Intervention services

- Prior to discharge, please enroll your patient in the Massachusetts Early Intervention Program.
- Read more details at www.mass.gov/dph/earlyintervention.
- To set up a meeting with Early Intervention, call 1-800-905-8437

If you have any feedback on this document, please contact
mghfcpatienteducation@partners.org.

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