





Supplemental Information

SUPPLEMENTAL FIGURE 1. Summary of Down syndrome-specific care.

| Action | Pre-natal | Birth up to 1 mo | 1 mo up to 1 yr | 1 yr up to 5 yr | 5 yr up to 12 yr | 12 yr up to 21 yr |
|--|-----------|--------------------------|--|---|---|--|
| 1. Confirm DS diagnosis with either CVS or amniocentesis prenatally or karyotype postnatally | | | | | | |
| 2. Review recurrence risk and offer the family referral to a clinical geneticist or genetic counselor. | | | | | | |
| 3. Offer parent-to-parent and support group information to the family. | | | | | | |
| 4. Use CDC DS-specific growth charts to monitor weight, length, weight-for-length, head circumference, or BMI. Use standard charts for BMI after age 10 years. | | All healthcare visits | | | | |
| 5. Order an echo, to be read by a pediatric cardiologist. | | | | | | |
| 6. Feeding assessment or video study if any: marked hypotonia, underweight (<5th %ile weight-for-length or BMI), slow feeding or choking with feeds, recurrent or persistent abnormal respiratory symptoms, desaturations with feeds | | Any visit | | | | |
| 7. Obtain objective hearing assessment (may be in NBS protocols) and follow EHDJ protocols. | | | Up to 6 mo | | | |
| 8. If TM can't be visualized, refer to otolaryngologist for exam with microscope until reliable TM and tympanometry exams are possible | | Every 3-6 mo | | | | |
| 9. Car safety seat evaluation before hospital discharge. | | | | | | |
| 10. CBC with differential | | By day 3 | | | | |
| 11. If TAM, make caregivers aware of risk/signs of leukemia (e.g., easy bruising/bleeding, recurrent fevers, bone pain) | | | | | | |
| 12. TSH | | At birth (if not in NBS) | Every 5-7 mo | Annually, and every 6 mo if antithyroid antibodies ever detected | | |
| 13. RSV prophylaxis based on AAP guidelines. | | Annually | | Through 2 yr | | |
| 14. Discuss cervical spine-positioning for procedures and atlantoaxial stability precautions. | | All HMV | | Biennially | | |
| 15. Assess for CAM use, discourage any unsafe CAM practices. | | All HMV | | | | |
| 16. Refer children to early intervention for speech, fine motor or gross motor therapy. | | Any visit | Up to 3 yr | | | |
| 17. If middle ear disease occurs, obtain developmentally-appropriate hearing evaluation. | | | When ear clear | After treatment | | |
| 18. Rescreen hearing with developmentally-appropriate methodology (BAER, behavioral, ear-specific). | | | Start at 6mo, every 6 mo until established normal bilaterally by ear-specific testing, then annually | | | |
| 19. Refer to ophthalmologist with experience and expertise in children with disabilities. | | | By 6 mo | | | |
| 20. CBC with differential if easy bruising or bleeding, recurrent fevers, or bone pain | | | Any visit | | | |
| 21. Assess for sleep-disordered breathing; if present, refer to physician with expertise in pediatric sleep disorders. | | | At least once by 6 mo, then all subsequent HMV thereafter | | | |
| 22. Ensure child is receiving developmental therapies, and family understands and is following therapy plan at home. | | All HMV | | | | |
| 23. CBC with differential and either (1) a combination of ferritin and CRP, or (2) a combination of serum iron and Total Iron Binding Capacity | | | | Annually | | |
| 24. If a child has sleep problems and a ferritin less than 50 mcg/L, the pediatrician may prescribe iron supplement. | | | | Any visit | | |
| 25. Vision screening | | | All HMV, use developmentally-appropriate criteria | Photoscreen (all HMV); if unable, refer to ophthalmologist annually | Photoscreen (all HMV); if unable, refer to ophthalmologist biennially | Visual acuity or photoscreening at all HMV, or ophthalmology-determined schedule |
| 26. If a child has myelopathic symptoms, obtain neutral C-spine plain films (see text for details). | | | | Any visit | | |
| 27. Obtain polysomnogram. | | | | Between 3-5 yr | | |
| 28. Prepare family for transition from early intervention to preschool. | | | | At 30 mo | | |
| 29. Discuss sexual exploitation risks. | | | | At least once | At least once | At least once |
| 30. Make developmentally-appropriate plans for menarche, contraception (advocate/offer LARC), and STI prevention. | | | | | As developmentally-appropriate, then all subsequent HMV | |
| 31. Discuss risk of DS if patient were to become pregnant. | | | | | At least once | At least once |
| 32. Assess for any developmental regression. | | | All HMV | | | |
| 33. Discuss and facilitate transitions: education, work, finance, guardianship, medical care, independent living | | | | | All HMV starting at 10 yr | |

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|  | Do once at this age | Abbreviations: DS, Down syndrome; CVS, Chorionic villus sampling; HMV, Health Maintenance Visit; BMI, Body mass index; CDC, Centers for Disease Control; EHDJ, Early Hearing Detection and Intervention; NBS, Newborn screen; CAM, Complementary and alternative medicine; BAER, Brainstem auditory evoked response; TM, Tympanic membrane; TAM: transient abnormal myelopoiesis |
|  | Do if not done previously | |
|  | Repeat at indicated intervals | |
|  | See report for end point | |