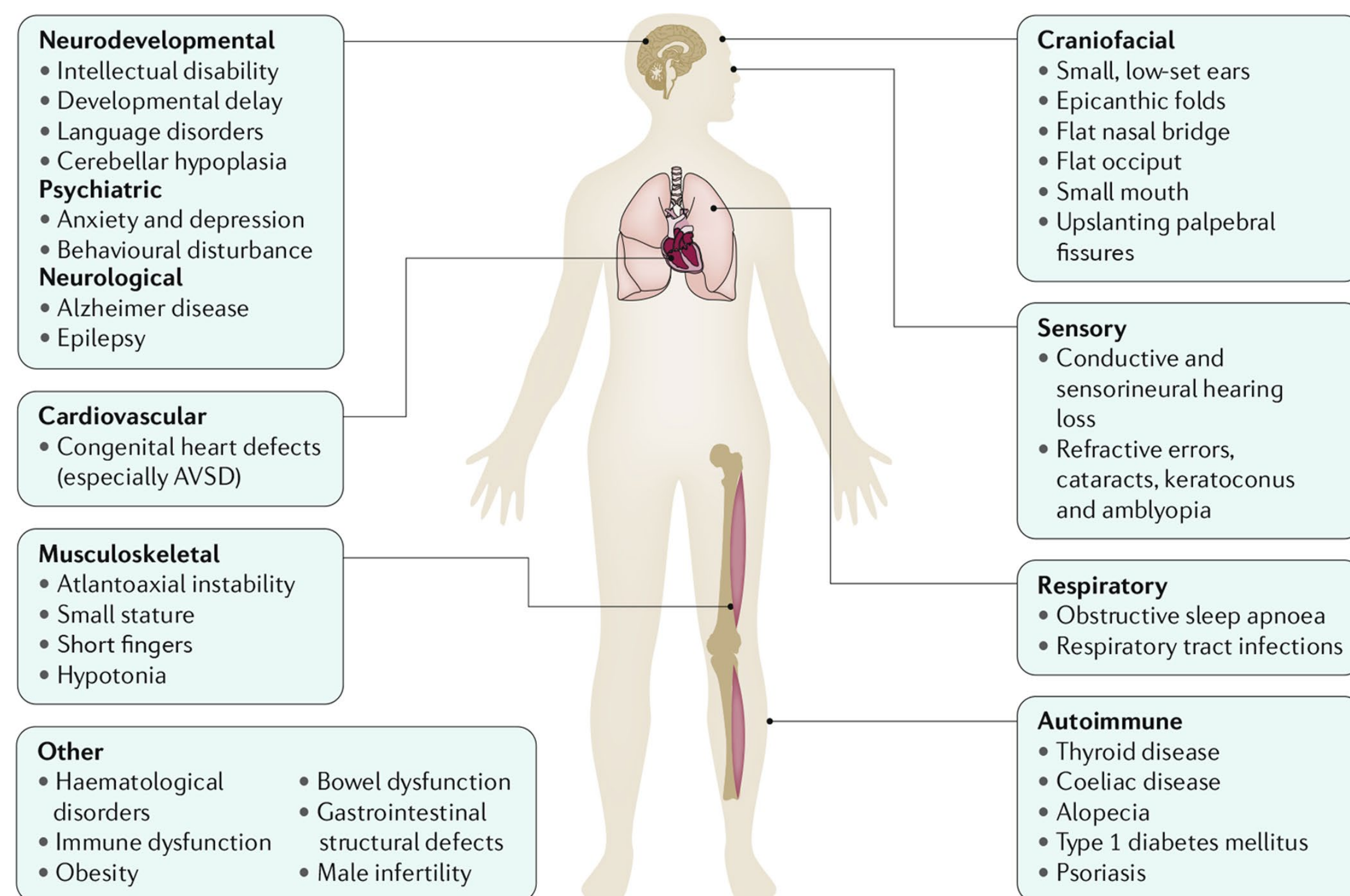


Introduction

- Down syndrome continues to be the most common chromosomal disorder in humans. Each year, about 6,000 babies are born with Down syndrome, which is about 1 in every 700 babies born.
- In Virginia it is estimated that about 123 babies are born annually with Down Syndrome.
- Down syndrome is associated with multiple co-morbidities that require frequent surveillance and early intervention



Method: Literature Review

- Keyword terms were entered into PubMed for years 2018-2023, yielding 169 results
- Terms included “Health supervision,” “children with Down syndrome,” “trisomy 21,” and “infant management.”
- Reviewed new AAP guidelines on health supervision for children with Down syndrome published May 2022

2011 Guidelines	2022 Guidelines
FISH testing and chromosome analysis	CVS, amniocentesis or karyotyping
Use standard WHO growth charts	Use CDC DS- Specific growth charts
CBC once any time from birth – 1 month	CBC with differential by day 3 of life
Refer to ophthalmology by 1 year	Refer to pediatric ophthalmologist by 6 months
TSH once from birth – 1 month, at 6 and 12 months, then annually	TSH at birth, every 5-7 months until 1 year of age, then annually or every 6 months if antibodies ever detected
Refer to early intervention at 1-3 year visit	Refer children to early intervention by 1 month
Discuss risk of respiratory infection	RSV prophylaxis
Assess for OSA symptoms by 1 year	Assess for sleep disordered breathing by 6 months
Discuss complementary & alternative medicine (CAM)	Assess for CAM use, discourage any unsafe CAM practices

Recommendations

- Confirm diagnosis with CVS or amniocentesis prenatally or karyotype postnatally
- Use CDC Down syndrome specific growth charts
- Have an echocardiogram read by cardiology
- Perform a feeding assessment or video study if any difficulties with feeds or desaturations
- Obtain objective hearing assessment
- CBC with differential
- Car seat trial before hospital discharge
- Refer to early intervention for SLP, PT, OT
- Discuss atlantoaxial instability precautions
- Draw and evaluation TSH level



Objectives

- To review current health supervision guidelines for infants born with trisomy 21
- To compare past AAP guidelines to new AAP guidelines
- To review the protocol at Sentara Martha Jefferson Hospital for infant’s born with Down syndrome
- To inform staff of SMJH of the recommendations from the AAP for neonates with Down syndrome
- To familiarize staff with a unique patient population

Education at SMJH

- No policies/protocols for infants born with trisomy 21 available at SMJH
- Nursing reference center links to academic journals and patient education handouts
- Reviewed AAP recommendations with SMJH nursing staff and contacted nurse manager

References

- Antonarakis, S. E., Skotko, B. G., Rafii, M. S., Strydom, A., Pape, S. E., Bianchi, D. W., Sherman, S. L., & Reeves, R. H. (2020). Down syndrome. *Nature reviews. Disease primers*, 6(1), 9. <https://doi.org/10.1038/s41572-019-0143-7>
- Bull, M. J., Trotter, T., Santoro, S. L., Christensen, C., Groat, R. W., COUNCIL ON GENETICS, Burke, L. W., Berry, S. A., Geleske, T. A., Holm, I., Hopkin, R. J., Introne, W. J., Lyons, M. J., Monteil, D. C., Scheuerle, A., Stoler, J. M., Vergano, S. A., Chen, E., Hamid, R., Downs, S. M., ... Spire, P. (2022). Health Supervision for Children and Adolescents With Down Syndrome. *Pediatrics*, 149(5), e2022057010. <https://doi.org/10.1542/peds.2022-057010>
- Martin, T., Smith, A., Breatnach, C. R., Kent, E., Shanahan, I., Boyle, M., Levy, P. T., Franklin, O., & El-Khuffash, A. (2018). Infants Born with Down Syndrome: Burden of Disease in the Early Neonatal Period. *The Journal of pediatrics*, 193, 21–26. <https://doi.org/10.1016/j.jpeds.2017.09.046>
- O’Neill, M. E., Ryan, A., Kwon, S., & Binns, H. J. (2018). Evaluation of Pediatrician Adherence to the American Academy of Pediatrics Health Supervision Guidelines for Down Syndrome. *American journal on intellectual and developmental disabilities*, 123(5), 387–398. <https://doi.org/10.1352/1944-7558-123.5.387>
- Seither, K., Tabbah, S., Tadesse, D. G., & Suhrie, K. R. (2021). Neonatal complications of Down syndrome and factors necessitating intensive care. *American journal of medical genetics. Part A*, 185(2), 336–343. <https://doi.org/10.1002/ajmg.a.61948>
- Telman, G., Sosnowska-Sienkiewicz, P., Strauss, E., Mazela, J., Marikowski, P., & Januszkiewicz-Lewandowska, D. (2022). Why Is Health Care for Children with Down Syndrome So Crucial from the First Days of Life? A Retrospective Cohort Study Emphasized Transient Abnormal Myelopoiesis (TAM) Syndrome at Three Centers. *International journal of environmental research and public health*, 19(15), 9774. <https://doi.org/10.3390/ijerph19159774>

Acknowledgements

The Blue Ridge Leadership Education in Neurodevelopmental Disabilities (Blue Ridge LEND) and this project is supported by the Health Resources and Services Administration (HRSA) of the U.S. Department of Health and Human Services (HHS) as part of an award totaling \$2,242,875. The contents are those of the author(s) and do not necessarily represent the official views of, nor an endorsement, by HRSA, HHS, or the U.S. Government. For more information, please visit HRSA.gov.