
Recommended Labs, Tests, and Procedures for Molly

(AAP = American Academy of Pediatrics)

- Ophthalmology Evaluation:** Experts recommend an ophthalmologic exam at least every two years for adults with Down syndrome, ages 21 and older.
- Celiac screen (total IgA & TTG-IgA).** Molly's caregiver indicated that within the past month of completing our intake, she had constipation that is hard to treat, nausea, passing excessive gas, bloating, mood swings, crying easily or for no reason. Molly has never had celiac testing done. Celiac disease is more common in people with Down syndrome.
- Sleep study.** Molly's caregiver indicated that within the past month of completing our intake, Molly experienced snoring, gasping, snorting, choking during sleep, sleeping on multiple pillows, not feeling refreshed despite adequate sleep, feeling down, depressed, or hopeless, crying easily or for no reason, moving slowly, distress about being alone. Molly has also never had a sleep study. Experts recommend that everyone with Down syndrome be evaluated for sleep apnea if symptoms arise. We would recommend a sleep study to assess for obstructive sleep apnea, which can occur in up to 75% of people with Down syndrome. Research has also shown that patients with Down syndrome may lose up to 9 IQ points within a year when their apnea is untreated.
- Thyroid function tests (TSH & FT4).** Molly's caregiver indicated that within the past 12 months of completing our intake, Molly has not had her thyroid levels drawn. Experts recommend that everyone with Down syndrome have their thyroid levels checked once annually, given the high incidence of thyroid disorders in patients with Down syndrome.

Potential New Conditions/Diagnoses to be Considered for Molly

We recommend that you consider initiating evaluations for the following conditions that can often co-occur with Down syndrome:

- Depression.** Molly's caregiver marked that within the past 6 months of completing our intake, Molly has experienced feeling down, depressed, or hopeless, little interest or pleasure in doing things, crying easily for no reason, moving slowly, distress about being alone. Generalized depression is a common co-occurring condition for people with Down syndrome. Resources and book recommendations are on our web page. Psychopharmacological consult should be considered if possible underlying medical causes can be eliminated.
- Arthropathies.** Molly's caregiver indicated that within the past month of completing our intake, Molly has experienced joint stiffness, joint pain. Gout and osteoarthritis are all common in people with Down syndrome, and a full standard work up should be pursued in patients presenting with symptoms of joint pathology, including a workup for other connective tissue disorders.
- Chronic constipation.** Molly's caregiver indicated that within the past month of completing our intake, Molly experienced constipation that is hard to treat. Chronic constipation is a common feature in patients with Down syndrome and can lead to behavioral issues if left untreated. Consider obtaining a KUB and trying medication to alleviate the constipation if needed.

Figure S5. Primary care provider (PCP) plan