

Finding New Friends
Down Syndrome

Pedicases

Objectives

- List the diagnostic features of Down syndrome and its associated complications.
- Describe the role of the clinician in management of Down syndrome, including: monitoring of growth/nutrition, recognition and prevention of medical complications, and participation in educational planning.
- Describe the psychosocial stresses faced by children with Down syndrome and their parents

Part I: Introduction

Jamie Peters is an 8-year-old girl who comes to your clinic with a complaint of headaches and neck pain. She was born at full term to a 28-year-old Gravida 2/Para 1 mother. Pregnancy was unremarkable, and birth was by normal spontaneous vaginal delivery. Apgar scores were 8 and 9. A pediatrician was called to the delivery room because the nurses noticed that Jamie had distinctive palpebral fissures, flat occiput, and transverse palmar crease on her left hand. Subsequent chromosomal analysis indicated that Jamie had trisomy 21 (Down syndrome).

Part I: Introduction

Her clinic record reveals that she had a small muscular ventricular septal defect that closed by the age of 18 months. She underwent surgical correction of duodenal atresia at age 3 days, and had bilateral tympanostomy tube placement for recurrent otitis media at age 3 years. Aside from intermittent bouts of constipation, she has been well since that time.

Developmentally, she spoke her first words at 17 months, walked at age 22 months, and completed toilet training at 4 1/2 years of age. She is currently in the third grade, in a mainstream classroom, and her mother reports she is able to "read at grade level."

Part I: Introduction

A full-time teacher's aide is assigned to Jamie and one other child. Jamie also receives occupational therapy once weekly and speech therapy twice weekly at school. Last year, she was quite happy and well behaved in the classroom. This year, however, she has demonstrated occasional school avoidance and noncompliant behavior. She becomes somewhat oppositional when asked to transition from a favorite activity to one that is less enjoyable. For example, when recess is over she will sit on the ground and refuse to go back to class.

She began to complain of headaches and neck pain three weeks ago. Her mother tells you, "*I think Jamie is complaining about these things so she won't have to go to school.*"

Part II: Next Steps

Jamie points to her head and over the right side of her neck when asked about the location of the pain. You ask, "Is the pain there all the time?"

"No."

"Does the pain go anywhere else on your body? Do your legs, arms or fingertips ever feel funny or tingly?"

"No."

"What time of the day is the pain the worst?"

"After lunch" she replies.

"Is there anything that makes the pain worse?"

"I don't know."

Part II: Next Steps

When asked about school, she responds that she likes her teachers and the classroom activities.

"Do you have friends in school?"

Jamie says, "Yes."

"Can you tell me the name of one of your friends?"

She is silent.

You turn to Jamie's mother and ask, *"Does she have any play dates outside of school?"*

Mom replies, *"She occasionally invites a younger girl from the neighborhood to come and play with her."*

Part II: Next Steps

Jamie's mother later tells you in private that while the younger neighbor occasionally does come over to play, she never invites Jamie to come to her house. She also lets you know that her schoolmates have never invited Jamie to any birthday parties or out-of-school activities.

Physical Exam:

Jamie is a warm and friendly 8-year-old with the typical facial features of Down syndrome. Her height is 126 cm. and her weight is 27 kg. Her body mass index is 17.0 (71 % on the NCHS growth chart). Her skin is dry, and she has mild atopic dermatitis. She wears glasses and has epicanthal folds and Brushfield spots on the outer third of the iris.

Part II: Next Steps

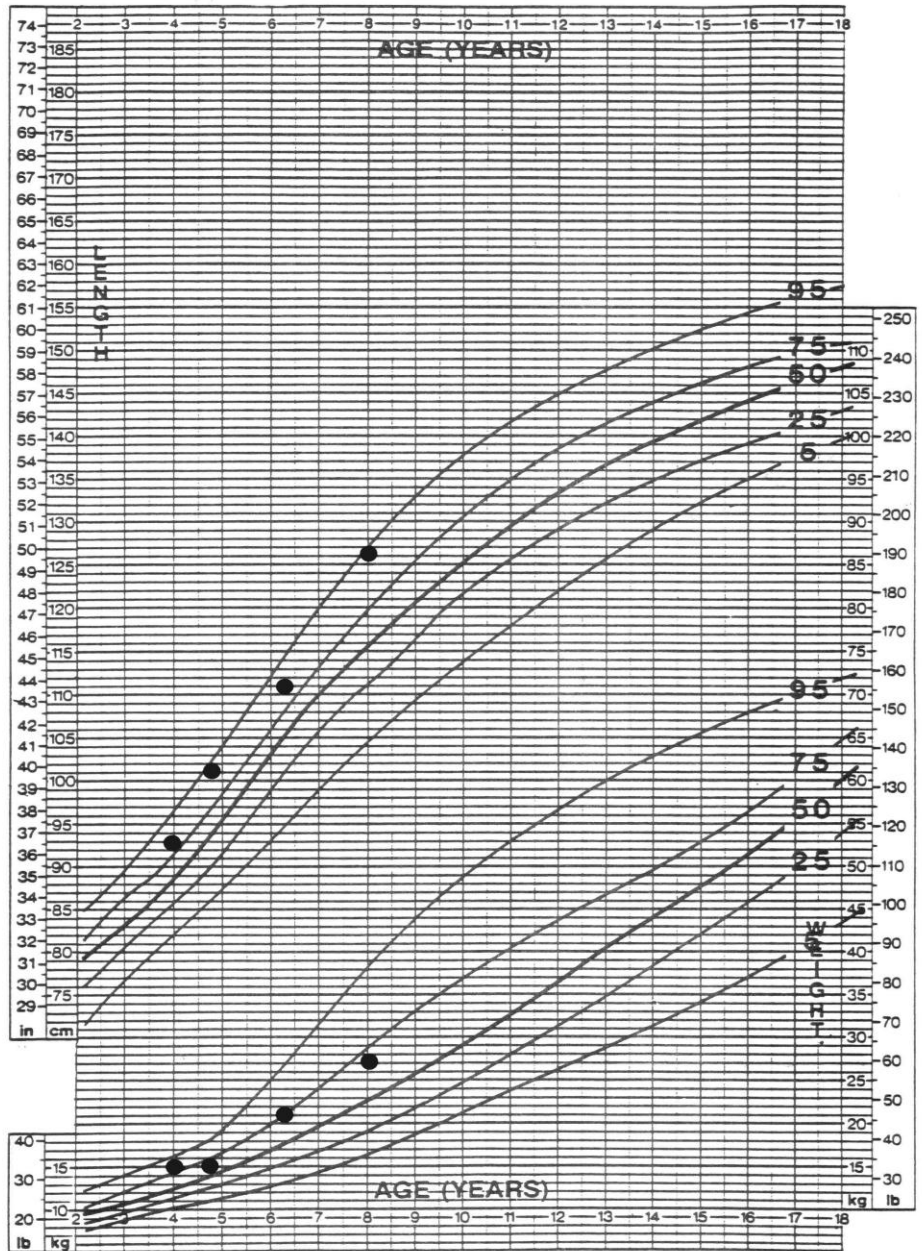
Physical exam continued:

She has an open bite with a tongue thrust. Her tympanic membranes are dull but not injected, and the motility is mildly reduced. Her thyroid gland is non-palpable.

Heart and lung sounds are normal. Abdominal exam reveals a midline surgical scar, and a moderate amount of palpable stool in the LLQ. Her breast and pubic hair are Tanner Stage I. You note a bilateral hallux valgus deformity. Her overall muscle tone is diminished with hyperextensibility of joints, and she has decreased truncal stability and rounding of the shoulders. There are no lateralizing neurological signs. Her gait is slightly immature although symmetric.

Girls with Down Syndrome: Physical Growth 2 to 18 Years

Girls with Down Syndrome:
Physical Growth: 2 to 18 Years



Results

- Lateral neck x-ray shows no evidence of occipito-atlanto or atlanto-axial instability.
- Her thyroid function tests are all normal.
- Audiometry reveals that her left ear is normal, and her right ear has mild low frequency conductive hearing loss. Her tympanogram is normal for the left, and shows negative middle ear pressure on the right.

Part III: Epilogue

Following her benign medical work-up, you reassured Jamie and her mother that there was no serious medical problem underlying her neck pain.

Following a consultation with a Behavioral Medicine specialist, Jamie's parents instituted a behavior management plan that included positive reinforcement (e.g., a sticker chart with subsequent reward) for staying in class and following teacher directions. Jamie continued to complain of pain in her head and neck. Her parents were initially reluctant to involve her in activities with other children who have developmental disabilities, as they feared that Jamie could learn additional negative behaviors.

Part III: Epilogue

They ultimately decided to try *Special Olympics*, which has on-going activities during the entire year. Jamie found acceptance there and had a wonderful time. She subsequently joined a dance class with typically developing peers at the local community center. Her complaints of head and neck pain have progressively diminished. She is not always compliant in school and the behavioral management plan needs to be reactivated from time to time.