

GROWTH AND DEVELOPMENT

Growth and Development, Case #1

Written by Linda O. Lewin, M.D.

A three-month-old full-term infant, who was 3000 g. at birth, now weighs 3420 g. Her height velocity has been normal. Her parents want to know if this is an adequate weight gain. What would you tell them? What are the common causes of poor weight gain at this age? How would you evaluate the infant?

Definitions for Specific Terms:

Height velocity- refers to the number of centimeters/year the patient has grown. In the first year of life the average growth velocity is 25 cm/yr but that varies from 38 cm/yr in the first 2 months to 12 cm/yr at 1 yr. At age 2 growth velocity is 10 cm/yr, from 2-4 yrs it is 7 cm/yr, from 4-5 yrs it is 6 cm/yr, and growth velocity is 5cm/yr from age 5 until the pubertal growth spurt.

Review of Important Concepts:

- Know the normal pattern of weight gain in the first three months of life
- List the common causes of poor weight gain in the first three months of life
- Describe the evaluation of a patient with poor weight gain at three months

Historical Points

This is an example of a situation in which a very detailed history is critical. These are some key points that can help rule in or rule out possible diagnoses:

- Other complaints about the baby, including sleeping too much, displaying unusual movements, spitting up, bowel problems, rashes, tiring easily, breathing hard, and other complaints that might point to a GI, cardiac, pulmonary, endocrine, neurologic, or infectious cause.
- Birth history looking for maternal illness, infection, medications, other exposures
- Family history of poor growth, feeding intolerance, inborn errors of metabolism, genetic syndromes
- Social history, including who lives with the baby, who feeds her, maternal depression or other illness
- Diet history, especially what the baby is eating. If breastfed then specific questions about how successful that has been and if formula fed then details about what formula, how it is mixed, and how much is being eaten.

Physical Examination Findings

The physical exam should also be detailed, looking for evidence of congenital anomalies, neurologic abnormalities, cardiac and respiratory problems, GI function, and general development.

1. General appearance:
Is the baby vigorous?

2. Vitals:
 - a. Tachycardia or tachypnea?
 - b. Fever?
 - c. Blood pressure abnormalities?
 - d. Low O₂ saturation?
3. HEENT:
Evidence of malformations of mouth, tongue, etc.
4. Lungs:
 - a. Respiratory distress?
 - b. Abnormal lung sounds?
5. Cardiac:
Murmurs, tachycardia?
6. Abdomen:
 - a. Large liver?
 - b. Other masses?
7. Neurologic:
 - a. Normal tone?
 - b. Reflexes?
 - c. Gag, suck, swallow?
8. Skin:
 - a. Rash?
 - b. Cyanosis?
 - c. Jaundice?
9. Lab work should be directed to the most likely causes based on the history and physical exam.

Clinical Reasoning

1. Is this adequate weight gain?

The average newborn loses up to 10% of his/her birth weight in the first few days of life, then is expected to be back at birth weight by 2 weeks of age. In the first 4 months, babies gain an average of 20-30gm (1oz) per day, from 4-6 months approximately 15-20gm/day, and from 6-12 months it is approximately 10-12gm/day. Most babies double their birth weights by 6 months.

Teaching suggestion: ask the learner(s) to calculate this babies' weight gain per day and determine if it falls within the expected guidelines. Can go to online growth chart and practice plotting this baby's growth at www.cdc.gov/growthcharts/.

The baby in the case has gained 420g since birth, or 14oz. We would expect in 90 days that the baby would have gained close to 2000g, so this is clearly not adequate.

2. What are the common causes of poor weight gain at this age?

Assuming that this is not a baby that was growing fine but then developed an acute illness that has caused her to lose weight, there is a long list of possible causes. They include:

a. Problems of intake:

- Too few calories with normal metabolic demands:
- Not enough given
- Improperly mixed formula
- Poor maternal milk supply due to poor diet, stress, medications (mother should continue prenatal vitamins while nursing)
- Improper use of other foods: most commonly water, juice
- Neurologic disease: if the baby has a poor suck reflex or uncoordinated swallow he/she might not get in enough. Those babies might choke/cough while feeding.
- Anatomic abnormality of the mouth (palate), pharynx, or esophagus
- Poor signaling of hunger: the baby doesn't cry when hungry or the parent doesn't recognize the baby's signal for wanting to eat

b. Enough calories but increased metabolic demands:

- Cardio-pulmonary disease: feeding is like exercise to babies and if they tire due to CHF or respiratory distress they may not take enough calories
- Chronic inflammation/infection
- Problems of usage of nutrients:
- Cow's milk and/or soy protein intolerance
- Significant GE reflux leading to excessive vomiting
- GI malformation leading to excessive vomiting
- Malabsorption of fats, carbohydrates, or proteins
- Metabolic disorder in which nutrients aren't used or leading to neurologic difficulties that cause poor feeding

Suggestions for Learning Activities:

- Present a number of scenarios in which the same patient presentation represents different underlying causes. Good examples are improper formula preparation, cow's milk intolerance (contrast it with lactose intolerance, which is extremely rare in newborns), congestive heart failure, and developmental delay, as they are more common than many of the other diagnoses. This could be presented in the opposite way as well and ask, for a given diagnosis, what would the learner expect to hear in the history and see in the physical exam and/or diagnostic studies?

Other Resources:

- <http://www.cdc.gov/growthcharts/>

Growth and Development, Case #2

Written by Linda O. Lewin, M.D.

A fifteen-month-old boy says no recognizable words. His parents are concerned and wonder if he needs speech therapy. How would you respond to their concerns? How would you evaluate this child?

Review of Important Concepts:

- Know the expected pattern of speech development in the first two years of life
- List at least one developmental screening tool that can help the clinician identify speech delay in young children
- List the common causes of speech delay in the first two years of life
- Describe the evaluation of a patient with speech delay at 15 months of age

Historical Points

- It is important to assess this child's development to date in all areas to determine if this is an isolated delay or if it represents part of a global developmental problem.
- It is also important to know if the child had some speech at one time but has regressed.
- Questions about hearing are also very relevant to a child without speech.
- Think of development in terms of gross motor (rolling, sitting, crawling, walking, running), fine motor (batting at objects, grabbing with whole hand, pincer grasp, scribbling, piling two objects), social (smiling, feeding self, pat-a-cake, waving, imitating adults), and speech (expressive language skills: cooing, babbling, one word, several words by 15 months and receptive language skills: following directions such as stop, come here, etc.).
- Ask if there is a family history of speech or other developmental delay, if the child has other medical problems or takes any medications, and what the social situation is like.

Physical Exam

The physical exam will be focused on the child's development in the four areas mentioned above, as well as looking for evidence of any systemic illnesses that might be related to developmental delay.

Clinical Reasoning

1. Is this a speech delay?
 - a. A 15 month old child should be able to say several words (in the 5-10 range) that are specific and recognizable to his close family members.
 - b. He should be able to point to common objects that others name, like his favorite toy, a person he lives with, a picture in a book, etc, and follow simple directions like "go get your cup."
 - c. A 15 month old that has no recognizable words is definitely delayed.
2. What is the expected pattern of speech development in the first 2 years?
 - a. By 3 months: turn to sounds, make cooing noises, cry differently for different needs, calm when spoken to
 - b. By 6 months: make gurgling sounds, "blow bubbles," make loud vowel sounds, respond differently to different tones in your voice, notice that some toys make sounds

- c. By 9 months: make repetitive consonant sounds like “ba ba ba” and “da da da” non-specifically, look in the direction of sounds, listen when someone talks to him/her
 - d. By 12 months: say one or two words that are specific, follow some very simple commands like “drink your milk,” point to something he/she wants, try to imitate words
 - e. By 18 months: Say 8-10 words that are specific, follow several simple instructions, point to objects, pictures, and people who are familiar when named, possibly point to body parts (must have been taught these)
 - f. By 24 months: say approximately 50 words, be at least 50% understandable to a stranger, put two words together in novel combinations, start to use pronouns like “mine,” follow simple commands without gestures
3. How do you respond to the child’s parents?
 - a. The clinician must tell the parents that the child is delayed, and that he/she is going to try to find out if there is a recognizable cause in order to decide how to proceed. This can be either easy or challenging, depending on the parents’ level of anxiety and the level of concern of the physician.
 - b. If the child seems to be globally delayed the discussion is much different than if there seems to be a simple expressive speech delay.
 4. What are some common screening tools to identify speech delay?

Commonly used tools are the Denver Developmental Screening Test, the Ages and Stages Questionnaire, the Child Development Inventory, and many others.
 5. What are the common causes of speech delay in a 15 month old?
 - a. Hearing impairment is the first thing to try to rule out when a child is speech delayed. Hopefully, this would have been found earlier than at 15 months either through screening in the newborn nursery or during well child visits. A hearing impaired child coos and makes normal noises early on, but does not develop babbling in the second half of the first year, and shows decreased or no response to sounds or music. Some causes to consider are chronic otitis media with effusion and exposure to ototoxic drugs. Children can be sent to the audiologist for formal testing if hearing impairment is suspected or if speech delay appears to be receptive as well as expressive.
 - b. Delayed speech can be associated with a global developmental delay, meaning that the child also shows delayed development in gross motor, fine motor, and social-adaptive development. There are a large number of causes of global delays, including prematurity (where the child might have to be corrected to gestational age), congenital infection, structural brain abnormalities, genetic syndromes, endocrine disorders, and others. Newborn screening with catch some of those problems, other testing should be done based on the information from a detailed birth history, family history, and past medical history.
 - c. Isolated expressive speech delay has a generally good prognosis. These are children who understand speech normally, but have trouble expressing themselves with speech. This is manifest as difficulty creating proper sentences, finding the right words, using grammar, and other processes of creating effective spoken communication. Poor pronunciation (phonological disorder) is a separate problem in which the child can put the words together correctly but can’t say them in an understandable manner. This is sometimes related to hearing impairment, with a potential history of exposure to ototoxic drugs or repeated bouts of otitis media in infancy.

6. How would you evaluate this patient?
- a. As noted above, the clinician first needs a detailed history, including prenatal and birth histories, family history, past medical history, exposure to ototoxic drugs, and developmental history to date.
 - b. Results of any previous testing, such as newborn hearing screening and serum newborn screening tests, should be reviewed.
 - c. The first decision point is whether this patient has a global developmental delay or an isolated speech delay.
 - If the problem is global, then work-up should follow from the clues gained from the history and physical exam.
 - If the speech delay is isolated, then hearing testing is appropriate, as well as a very good ear exam looking for fluid in the middle ear or scarring from repeated otitis media.
 - Referral to the state's Infants and Toddler's program, or its equivalent, is appropriate for evaluation and ongoing therapy as necessary.

Other Resources:

- US Preventive Services Task Force: Screening for Speech and Language Delay in Preschool Children. At: <http://www.uspreventiveservicestaskforce.org/uspstf/uspshdv.htm>.
- Agency for Healthcare Research and Quality: Screening for speech and language delay in preschool children: recommendation statement. At: <http://www.ngc.gov/content.aspx?id=8396>.
- Leung A, Kao CP. Evaluation and Treatment of the Child with Speech Delay. 1999. <http://www.aafp.org/afp/990600ap/3121.html>

Growth and Development, Case # 9

Written by Kirsten Hawkins, M.D.

A 14 year-old female has not started her periods. What important information should you gather from the history and physical examination?

Definitions for Specific Terms:

Primary Amenorrhea- Is defined as the lack of menses by age 14 in the absence of normal development of breasts and pubic hair or by the absence of menstruation by age 16 in the presence of normal development of breasts and pubic hair.

Sexual Maturity Rating. (a.k.a.: Tanner Staging)- There are five distinct pubertal stages as developed by Marshall and Tanner.

Thelarche- Onset of breast development or breast budding

Review of Important Concepts:

Historical Points

- Medical History: Menarche usually occurs in Breast Stage 3 or 4 and approximately 3.3 years after their growth spurt and 2.3 years after breast development. The normal range for menarche is 9-15 years with an average age of approximately 12.5 years. 95-97% of females reach menarche by age 16 and 98% by 18 years.
- Past Medical History: Patients with a history of cancer, autoimmune disease, solid organ transplantation, and chronic disease are at risk for primary amenorrhea. A history of any CNS, ocular, olfactory, pelvic, gonadal, and genital abnormalities should be obtained. Growth parameters should be graphed. Family history including parental age of pubertal development is needed.
- Confidentiality: Adolescent patients may be unwilling to share certain information when their parents are present. It's important to not only take a history with the patient's parent/guardian in the room, but also to discuss certain topics privately with the patient. Typically, after the initial history is taken, parents should be asked to step out for a confidential discussion. In addition, the patient should be assured of confidentiality, with the exception of cases where immediate harm may come to the patient or to another person.
- Sexual History: A detailed sexual history is also very important in this case.

Physical Exam Findings

1. A careful physical examination is important, including height and weight, blood pressure, palpation of the thyroid gland, and Tanner staging of the breast and pubic hair. Facial defects may be associated with hypothalamic-pituitary dysfunction. Renal and vertebral anomalies and hernias are associated with mullerian malformations.
2. A careful gynecologic examination is important in identifying the cause of primary amenorrhea.
 - a. The breast examination may reveal small, pale areolae as a sign of chronic low estrogen.

- b. If amenorrhea is present with galactorrhea, consider a prolactin-producing pituitary adenoma.
 - c. The external genitalia should be examined, looking for an enlarged clitoris, an abnormal hymen, or evidence of estrogen deficiency (thin, pink, vulvar mucosa).
3. Vaginal patency should be established with moistened cotton or Dacron tipped applicator or a moistened gloved finger. If a bimanual exam is possible, look for a mass or ovarian enlargement. A rectal exam may be necessary to help establish the presence of a cervix or to find a mass proximal to an obstructed vaginal canal.
 4. The presence of sparse or absent pubic hair with full breast development suggests androgen insensitivity syndrome.
 5. Identify hirsutism, severe acne, clitoromegaly, and scalp hair thinning as signs of hyperandrogenism.
 6. Evaluate the patient's nutritional status and plot the growth parameters.

Clinical Reasoning

1. Generate a differential diagnosis for this patient.
 - a. Turner's Syndrome
 - b. Kallman's Syndrome
 - c. Constitutional delay of growth
 - d. Celiac Disease
 - e. IBD
 - f. Cystic Fibrosis
 - g. Pituitary failure
 - h. Gonadal failure
 - i. Eating disorders
 - j. Pituitary adenoma
 - k. Craniopharyngioma
 - l. PCOS
 - m. Mayer-Rokitiansky-Kuster-Hauser Syndrome
 - n. Androgen Insensitivity
 - o. Pregnancy
 - p. Thyroid disorders
2. What lab tests might you order as an adjunct to your history and physical exam? Depending on your history and exam you might need:
 - a. Karyotype
 - b. complete blood count
 - c. erythrocyte sedimentation rate
 - d. Celiac evaluation
 - e. electrolytes
 - f. blood urea nitrogen
 - g. creatinine
 - h. glucose
 - i. calcium

- j. phosphorus
 - k. albumin
 - l. liver enzymes
 - m. urine analysis
 - n. FSH
 - o. TSH
 - p. Free T4
 - q. Prolactin
 - r. testosterone
 - s. DHEAS
 - t. Regardless of reported sexual activity, the patient should have a urine or serum human chorionic gonadotropin (HCG) test
3. What radiology study might you obtain as an adjunct to your physical exam?
- a. A pelvic ultrasound or pelvic MRI may be helpful to identify structural lesions, intrabdominal testes, or congenital malformations.
 - b. A head MRI may identify a pituitary adenoma or craniopharngioma.

Suggestions for Learning Activities:

Ask the students to perform a role play for this scenario. Students should practice asking the “parent/guardian” to leave the room, taking a detailed social history from the adolescent “patient.”

Other Resources:

- Nelson. L, Primary Ovarian Insufficiency N Engl J Med 2009; 360:606-14.
- Neinstein, et al. Adolescent Health Care- A Practical Guide. 5th Ed. 2008
- Sanfillipo, et al. Pediatric and Adolescent Gynecology, 2nd Edition, 2001
- Emans SJ, Laufer MR, Goldstein DP. Pediatric and Adolescent Gynecology, 5th Edition, 2005