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PEDIATRIC STONE DISEASE

TRENDS // TRIGGERS // TREATMENT

Winter sports injuries

Prenatal diet linked to ADHD

ANTIBIOTICS AND ALLERGIES
Our mission: Office- and hospital-based pediatricians and nurse practitioners use Contemporary Pediatrics’ timely, trusted, and practical information to enhance their day-to-day care of children. We advance pediatric providers’ professional development through in-depth, peer-reviewed clinical and practice management articles, case studies, and news and trends coverage.

Dr. Jellinek, with Michael Murphy, EdD, developed the Pediatric Symptom Checklist, the most widely used, brief screening instrument to identify children with emotional problems.

I am 68 years old, and I retired 2 years ago because I was totally burned out. I was a full-time neonatologist enjoying my work every day and doing what I thought was a good job. I am a sucker for numbers, so when I was hired to start a new intensive care unit in Nashville, I set up a database and kept track of all my patients and outcomes. [My practice] started with 2 full-time people and ended 30 years later with 6. I was in practice for 40 years, the last 30 spent running this NICU and loving every minute.

Well, I never knew burnout until about 10 years ago. I had always overseen the practice and acted as an administrator [of] the financial side of it. For years we charged what we thought was right and insurance companies payed us well. Then things started to change. We had to have contracts with each insurance company and allow discounts, which increased our prices. Then insurance companies started telling us how to write our notes to “justify” our charges and fulfill codes. The notes grew in length but very little in content.

At the same time, our nurses became “partners” and our patients “clients,” and hospital administration began to demand things their way. [T]hings got so out of hand that . . . we sold our practice to a corporation that took over billing and collections. They increased our charges and demanded we use their EMR, which generated 2 to 3 page notes every day that said nothing but complied with the insurance requirements.

“Medicine has been chained.”

[Next], the government [got] involved in our practice, telling us what we should do, how to treat our patients, and what standard of care was. Eventually the hospital demanded we order everything through the computer. Nurses’ notes disappeared and everything became electronic. We started to spend more time with the computer than the patient, and lost the notion of what things cost. We just saw patients and [charted]. Then I quit.

Medicine is a clinical science that has been chained . . . . We have become doers rather than thinkers. Even putting hands on a patient has now been substituted by monitors and machines.

This is not why people go into medicine. Patient interaction, touch, and communication [are] what medicine was all about. The doctors of today do not have much of that anymore. They have become computer technicians [and] protocol followers. That is why people get burned out!

Jorge Rojas, MD  
Nashville, TN

Dr. Schuman responds: Thanks for your comments, and I hope you’re enjoying your well-deserved retirement. Most pediatricians continue to enjoy caring for children but despise the atmosphere in which we practice today because, as you note, it has de-personalized medical care. Nearly 1 in 2 physicians is already “burned out”! Because we are the first-line providers of care, our voices should be listened to, but we protest individually, not collectively. Unfortunately, the AAP advocates for our patients, not for its physician members. The only way forward may be for physicians to unionize so we can collectively work toward improving medical care for patients as well as our own work environment. If we staged strategic actions as a group that would not jeopardize patient care, I think we would get the attention of government bureaucrats and insurance companies rather quickly!

Andrew J Schuman, MD  
Section Editor, Peds v2.0  
CEO, MedGizmos.com
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congressional fact of life is the dangerous “Christmas tree” bill, which passes with so much on it that many things don’t get the scrutiny they deserve.

Thus, the 21st Century Cures Act, signed into law by President Obama on December 13, 2016, is the equivalent of maybe 500 regular-sized pages that modify the US Food and Drug Administration (FDA) drug and device approval process, fund precision medicine and a cancer moon shot, and make numerous changes to Medicare, to name some examples.

In a subtitle on vaccines, the Cures Act calls for a report from the US Department of Health and Human Services on promoting innovation in vaccine development, and it updates the vaccine injury compensation system so that when a pregnant woman receives a covered vaccine, the child in utero will be considered covered by the system. It also has specific mandates for the Advisory Committee on Immunization Practices (ACIP) to, as appropriate, consider a vaccine at its next regularly scheduled meeting after the vaccine is licensed by the FDA. If ACIP does not make a recommendation, it must provide an update on its review.

The law mandates that the Centers for Disease Control and Prevention review the ACIP’s recommendation process, “including the identification of any areas for which flexibility . . . is necessary and the reason for flexibility,” and the extent to which the processes used by its work groups are consistent with the other work groups.

Walter A. Orenstein, MD, former director of the US Immunization Program and now director of Vaccine Policy and Development at Emory University, Atlanta, Georgia, commented on the provisions, saying, “I think there clearly are some misunderstandings of what the processes are and how we try and make our immunization schedule as effective and safe as it could be.”

The Cures Act also contains major legislation to expand mental health work, including creating the position of assistant secretary for mental health and substance abuse. In addition, it mandates that the National Institutes of Health help create a research network on pediatric rare diseases or birth defects, and it continues the voucher program within the FDA that gives drug sponsors incentives to develop new drugs for rare pediatric diseases.

The MACRA system is named for the 2015 bipartisan legislation “Medicare Access and CHIP Reauthorization Act,” which created the tsunami of change through Medicare, and basically just extended funding for the Children’s Health Insurance Program (CHIP) through fiscal year 2017.

The Centers for Medicare and Medicaid Services (CMS) says MACRA’s “Alternative Payment Models” (APM) rules, developed with the clinician community, add incentives for high-quality and cost-efficient care: They can apply to “a
specific clinical condition, a care episode, or a population.”

Deeper in the system’s second model of payment, the “Advanced Alternative Payment Models,” there is a multipayer component, explains Lindsey Browning, MPP, program director at the National Association of Medicaid Directors. In the out years, “If a provider is participating in Medicare APMs as well as participating in other payors’ APMs, it may help them receive that Medicare bonus under the program,” she says.

One situation like this, Browning points out, would be where a pediatrician is part of a hospital system. That means a 5% Medicare Part B bonus for having a certain number of patients or percent of payments through the Advanced APMs. It is an indirect influence on some of the payment system reform happening in Medicaid, she says.

Another part of MACRA will be an effort to increase electronic health records use, with systems pushing providers to meet the requirements. Also, in general, Medicare is such a huge payer that it has a big influence on other payers.

In a June comment on what was then the draft rule, the American Academy of Pediatrics (AAP) said pediatrics would be most clearly impacted through the rules’ APM pathway as a Medicaid medical home. However, the AAP said, the CMS had provided an extremely limited opportunity for that. Such a Medicaid medical home can only qualify for the 5% APM payment if information technology is meaningfully used; quality structures similar to the Merit-Based Incentive Payment are implemented; and the practice is comparable to the officially defined medical home or bears some degree of risk. Those are highly problematic for pediatrics, the comments said.

Browning says as the system continues to evolve, the question is how to move toward value-based purchasing while linking payment to quality measures that make sense for kids, adults, individuals with persistent mental illness, and others.

The CMS published the final rule, the equivalent of perhaps 1000 standard pages, in the Federal Register on November 4, but added another comment period that ended on December 19, 2016.

Reversal of MACRA is considered unlikely under the Trump administration, given the bipartisan support the legislation had.

**Trump nominates new leader for CMS**

President-elect Trump has announced he will nominate Seema Verma, MPH, a consultant with extensive work in Medicaid in several states, to lead the Centers for Medicare and Medicaid Services (CMS).

The head of the consulting firm SVC Incorporated, Indianapolis, Indiana, Verma was called by the Trump team the architect of Indiana’s Medicaid expansion, Healthy Indiana Plan (HIP), under the governors Mitch Daniels (R) and Mike Pence (R).

The SVC website says Verma has worked on Medicaid, insurance, and public health with governors’ offices, state Medicaid agencies, health departments, and departments of insurance. She and her firm, says the site, among other things developed Medicaid reform programs including waivers for Iowa, Ohio, and Kentucky, and helped in Tennessee’s coverage expansion proposal and Michigan’s Section 1115 Medicaid waiver.

Verma is best known for the Healthy Indiana Plan, approved by the CMS in 2015, and its requirement that participants must contribute a small amount. If participants above the poverty line begin paying the fee but then stop, they can be disqualified for 6 months. In a July 2016 report, the Lewin Group, Falls Church, Virginia, found that of the Indiana residents who could be disqualified, 16% always worried about affording their payment and 29% worried usually or sometimes.

After meeting with Verma, Senate Finance Committee Chairman Orrin Hatch (R-Utah) stated middle-class families have struggled to find affordable care under Obamacare, **CONTINUED ON PAGE 32**
Medication or placebo to prevent migraine?

A trial comparing amitriptyline, topiramate, and placebo for prevention of migraine in children and adolescents with a history of migraine found that all 3 had about the same effect on reducing headache frequency or headache-related disability. The active drugs were associated with higher rates of adverse events versus placebo.

The 328 participants in the trial were drawn from 31 US sites. Aged from 8 to 17 years, the youngsters had a diagnosis of migraine with or without aura or chronic migraine without continuous headache, and had experienced headaches 4 or more days during a baseline period of 28 days. After this baseline period, investigators assigned participants to oral amitriptyline, topiramate, or placebo in a divided dose of 1 capsule twice daily.

The target dose was 1 mg/kg of body weight per day for amitriptyline and 2 mg/kg per day for topiramate. The dose was escalated every 2 weeks for 8 weeks followed by a 16-week constant-dose phase of the highest dosage achieved. The 24-week treatment period was followed by a 2-week weaning period and a 4-week follow-up. Participants completed a daily headache diary.

At the end of the trial, investigators tallied how many children in each group achieved a reduction of 50% or more in the number of days on which they had a headache during the final 28 days of treatment compared with the number of days on which they had a headache in the 28-day baseline period. Differences among the groups were insignificant: 52% in the amitriptyline group reached the 50% reduction goal compared with 55% of the topiramate group and 61% of the placebo group. Nor were any significant differences seen among groups in headache-related disability, the number of headache days, or the proportion of patients who completed the 24-week treatment period.

Patients who received amitriptyline or topiramate had higher rates of adverse events than those receiving placebo. For the amitriptyline group, these adverse events included fatigue (30% vs 14% in the placebo group) and dry mouth (25% vs 12%, respectively), and in the topiramate group, paresthesia (31% vs 8%) and weight loss (8% vs 0%) (Powers SW, et al. N Engl J Med. October 27, 2016).

Effective early autism intervention has long-term impact

A follow-up trial conducted 6 years after the conclusion of a randomized controlled trial of early intervention in autism spectrum disorder demonstrated that the intervention had a long-term effect on autism symptoms and continued effects on parent and child social interaction.

The intervention, the Preschool Autism Communication Trial (PACT), was conducted in 3 specialized clinical services centers in the United Kingdom in children aged 2 to 4 years with so-called core autism. About half of participants were assigned to the PACT intervention and the other half to usual treatment. The PACT is a developmentally
targeted social communication program that focuses on optimizing parent interactive behaviors to improve child communication and more general autism symptoms. It provides 12, 2-hour therapy sessions during a 6-month period followed by monthly support sessions for a further 6 months. In addition, parents spend 20 to 30 minutes a day performing planned practice activities with the child.

Evaluations of 80% of the original 152 PACT participants 6 years after the conclusion of the PACT trial, when they had a mean age of 10.5 years, found that although the proportion of participants with high severity symptoms had increased in both groups, the PACT group fared better than the usual treatment group. Specifically, 29% in the PACT group had high symptom severity scores compared with 44% of the usual treatment group. In addition, parental reports also indicated that PACT had a treatment effect on symptoms as well as social communication skills (Pickles A, et al. Lancet. 2016; 388[10059]:2501-2509).

my take
This study provides strong, evidence-based support for the American Academy of Pediatrics (AAP) recommendation for routine screening for autism. Routine screening leads to early diagnosis and treatment, which in turn lead to an improved prognosis. The AAP continues to recommend screening for autism at ages 18 and 24 months and whenever the family expresses concern about the diagnosis (Pediatrics. 2007;120[5]:1183-1215). —Michael G Burke, MD

Overweight is not treated in children hospitalized with asthma
A retrospective record review for youngsters admitted with asthma to a Midwestern children’s hospital revealed that overweight and obesity were underrecognized, underdiagnosed, and undertreated.

Review of the children’s records showed that providers did not document body mass index (BMI) for 96.7% of the total sample of 510 patients aged 3 to 17 years, of whom 19.6% were obese and 13.3% were overweight.

Providers supplied a discharge diagnosis reflecting overweight or obesity in addition to the primary diagnosis of asthma for only 9 of the 168 patients with overweight or obesity, and all 9 of these youngsters were obese. Similarly, all 14 of the 168 overweight/obese asthmatics who received treatment for their weight were obese (Borgmeyer A, et al. Hosp Pediatr. 2016;6[11]:667-676).

my take
In the community hospital where I work, most hospitalized children have a short length of stay. However, even a short hospitalization is many times longer than a primary care office visit, and we should use that time in the hospital to address health issues other than the reason for admission. These authors identify 1 frequently missed opportunity. Others include resolving immunization delay, especially promoting influenza vaccination; screening for food insecurity; and enhancing education on chronic health conditions and preventive care. —Michael G Burke, MD

also of note
In newer urinalysis techniques, concentration matters. In a retrospective study of 27,000 infants aged younger than 3 months evaluated for urinary tract infection (UTI) with paired urinalysis and urine culture, investigators set out to determine the optimal urine white blood cell (WBC) threshold for UTI when using an automated urinalysis system stratified by urine concentration. Based on their analysis of the data, the authors recommend using pyuria thresholds of 3 WBC/high-power field (HPF) in dilute urine (specific gravity, <1.015) and 6 WBC/HPF in concentrated urine (specific gravity, >1.015) for a presumptive diagnosis of UTI. They also note that positive leukocyte esterase by automated dipstick is a strong indicator of UTI (Chaudhari PP, et al. Pediatrics. 2016;138[5]:e20162370).
As the prevalence of stone disease has grown, so has the importance of understanding disease process, diagnosis, and management.

IRENE M MCALEER, MD, JD, MBA

Urolithiasis occurrence is increasing in both adults and children in the United States, with nearly 1 in 11 adults having a stone at some time in their life. Unfortunately, stone occurrence in children also appears to have increased from 1% to 2% in the 1950s to 1970s to almost 10%, where previously the rate of stone incidence was only 18 per 100,000 in 1999 to 57 per 100,000 in 2008. Adolescent girls (aged 12 to 18 years) have a higher rate of occurrence than the other groups studied, although the overall sex distribution in all age ranges was about the same.

Hospital admission rates for urolithiasis increased to 1 in 683 admissions in the 2002 to 2007 time period.

With more children and adolescents developing kidney and ureteral stones, it is critical that the pediatric care community (pediatricians, nurse practitioners, primary care providers, and family medicine physicians) understand how to evaluate, treat, and prevent recurrence of stones in their patients. This article provides background on the risk factors for stone disease, its presentation in children, and the changes in diet that increase that risk, and offers practical tips on the evaluation, treatment, and prevention of stones.

$229 million
ESTIMATED ANNUAL ECONOMIC BURDEN FOR US HOSPITAL ADMISSIONS RELATED TO PEDIATRIC NEPHROLITHIASIS

AND $146 m/yr FOR ER ENCOUNTERS

Risk factors
Pediatric stone disease has different geographic and racial prevalence rates. Stones are very common in children in the Middle East, Pakistan, India, and Southeast Asia. Children in developing countries tend to
have more bladder calculi than calculi elsewhere in the urinary tract. Bladder stone composition in these children consists predominantly of ammonium acid, uric acid, and urate, likely because of the relatively low availability of dietary phosphate in these countries.\(^2\)

Children of African descent worldwide rarely have stones, whereas in the United States, Caucasian children are more likely to suffer from urolithiasis, especially if they are from the Southeast region. Stones are more likely to be found in the kidneys and ureters than in the bladder in American children.

Previously, most children who developed kidney stones also had anatomic abnormalities that increased their likelihood to develop stones, such as obstruction of the ureter or renal pelvis, exstrophy, or static drainage with horseshoe kidney or megaureter. Now, between 40% and 50% of children with urolithiasis have metabolic abnormalities identified, whereas only 30% of stones are associated with genitourinary abnormalities. Most likely, the children with anatomic abnormalities and urolithiasis have concomitant metabolic risk factors.

**DIET**

Much of the increased incidence in stone formation in children and adolescents is attributed to major dietary changes in the United States over the past few years. The Centers for Disease Control and Prevention (CDC) recently published that the prevalence of obesity in children aged 2 to 19 years is about 17%, affecting about 12.7 million children and adolescents. The prevalence is particularly high in Hispanics (21.9%) and non-Hispanic blacks (19.5%), while it is only 14.7% in non-Hispanic white children.\(^3\)

Children, particularly adolescents, are not drinking as much water or milk as they did previously. Increased consumption of sugary drinks has added to the increased obesity rates in children. Increased

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**4 MAJOR TYPES OF KIDNEY STONES OCCUR IN CHILDREN**

**CALCIUM STONES** are the most common type of kidney stone occurring in 2 major forms: calcium oxalate and calcium phosphate. Calcium oxalate stones are more common. Calcium oxalate stone formation has various causes, including high calcium excretion, high oxalate excretion, or acidic urine. Calcium phosphate stones are caused by alkaline urine.

**URIC ACID STONES** form when the urine is persistently acidic. A diet rich in purines—substances found in animal proteins such as meats, fish, and shellfish—may cause uric acid. If uric acid becomes concentrated in the urine, it can settle and form a stone by itself or along with calcium.

**STRUVITE STONES** result from kidney infections. Eliminating infected stones from the urinary tract and staying infection free can prevent more struvite stones.

**CYSTINE STONES** result from a genetic disorder that causes cystine to leak through the kidneys and into the urine in high concentration, forming crystals that tend to accumulate into stones.

Source: NIH’s National Institute of Diabetes and Digestive and Kidney Diseases.
sodium consumption through processed foods also has increased stone formation through increased urine calcium excretion. The most common abnormalities found have been hypercalcuria and hypocitraturia. Other metabolic problems seen, but less frequently, in children are hyperoxaluria, cystinuria, and hyperuricosuria. The most common stones found in US children are calcium oxalate (40% to 65% of all stones), calcium phosphate (14% to 30%), magnesium ammonium phosphate (struvite, 10% to 20%), cystine (5% to 10%), and uric acid (only 1% to 4%). In children, increased uric acid in urine promotes calcium oxalate stone formation, whereas uric acid stones are more commonly seen in adults.

Stones form when there is a supersaturation of these minerals. Promotion of crystallization through low total urine volume, increased concentrations of stone-forming ions, and decreased concentrations of inhibitors of crystallization all have been implicated in the increased rate of stones in children. As mentioned previously, children notoriously fail to have adequate fluid intake, particularly water intake. They also are more likely to drink beverages that increase their risk of stone formation, such as dark, caffeinated, sugary sodas. These trends have been increasing over the past 10 years, as have “fad diets” that may increase risk of stone formation when used in combination with low-volume or high-sugar-content fluid intake.

Low-carbohydrate or high-protein diets or vegan diets that may be high in oxalate intake are becoming more prevalent in children and adolescents. Vegan diets also increase the risk of hyperuricemia and hyperoxaluria from increased intake of foods high in oxalates (such as kale, spinach, and rhubarb). Many of these diets restrict intake of milk or dairy products necessary to keep adequate calcium intake that allows binding of calcium to ingested oxalates, as well as being high in salt intake. The fruits and vegetables eaten in such diets are the main sources of dietary oxalates. These diets contribute to a high dietary load of oxalates and, if calcium is Restricted, will actually increase risk of calcium oxalate stone formation.4

In the past, by contrast, high milk intake helped children maintain adequate calcium intake to prevent stones. Appropriate calcium intake decreases the absorption of oxalate in the intestines, preventing increased oxalate excretion by the kidneys that potentiates stone formation.

**SODIUM INTAKE**

Increased sodium intake by children in the United States is also rising above recommended dietary allowances. This increase in body sodium increases the excretion of urinary calcium, which promotes supersaturation of calcium and subsequent stone formation.2,5

**OBESITY**

Increased obesity in children also may be putting them at risk for stone formation, as is being seen in adults. Obesity causes a decrease in urine pH and increased excretion of sodium, phosphorous, and oxalate, increasing the rate of stone formation. As the rate of childhood obesity tripled from 1980 to 2002, obesity as a cause of increased stone rate in children has not been as well founded as it has been in adults.3

**HORMONES**

Increases in stone occurrence in adolescent girls may be hormonally driven by estrogen increases with the onset of puberty. Similarly, it has been found that hormone replacement therapy, particularly using estrogen supplements, in postmenopausal women causes a decrease in calcium excretion and an increase in citrate secretion. However, postmenopausal women have an increased stone rate over premenopausal women possibly from...
increased calcium oxalate supersaturation seen in the estrogen supplemented group. Thus, increased estrogen levels in adolescent girls and postmenopausal women on supplements may cause higher stone prevalence attributed to increased calcium oxalate supersaturation.

SEIZURE MEDICATIONS

Additionally, children with seizure disorders who are placed on a ketogenic diet or high-protein diet to prevent seizures have a higher risk of stone formation. The high-protein intake in these diets may raise urinary oxalate excretion increasing the likelihood of urinary lithogenesis. In this population, antiseizure medications (topiramate and zonisamide) are known to potentiate stone formation by increasing hypocitraturia. Also, these children frequently are fluid restricted or cannot adequately hydrate themselves to offset citrate loss.

Stone presentation in older children is similar to adult stone presentation, commonly flank pain, abdominal pain, nausea, and vomiting. Younger children do not always present this way. In fact, only 10% to 14% of younger children present with typical acute renal colic symptoms. These children tend to have vague symptoms and less localized pain but may present with hematuria or urinary tract infection.

Diagnosis, treatment, and prevention

Although many children and adolescents with urolithiasis present with abdominal or colicky flank pain, similar to adults, many children may have nonspecific symptoms, such as generalized abdominal pain, nausea, vomiting, or nonspecific findings consistent with urinary tract infection. Many children will have gross hematuria as initial presenting sign of urolithiasis.

Treatment of children with renal stones is actually similar to that of adults. Many children will pass similarly sized stones as adults. Fifty percent of children will pass stones 4 mm to 5 mm in size regardless of the child’s size. Increased hydration and pain management with either nonsteroidal anti-inflammatory drugs (NSAIDs) or, rarely, narcotics help children manage their symptoms while passing a stone. Many children can be treated as outpatients and do not require hospital admission on presentation or when symptoms are controllable.

Alpha-adrenergic blockade (tamsulosin) has been used in children as well. The use of stone expulsion treatment particularly to facilitate passage of distal ureteral stones appears to have similar success in children as in adults and the medications are well tolerated by children, although current studies are limited.

Long-term prevention includes increased fluid intake, particularly water, up to 2 liters per day to
2.5 L/d in adolescents, or more in cystine stone formers. Improved fluid intake and salt and sugary drink limitations are critical for prevention of recurrence of stones in most children and adolescents, but are difficult to achieve, just as these improvements are similarly difficult to achieve in adults.

Specific treatments depend, if possible, on obtaining urine and serum testing for causative factors in stone formation. This is somewhat difficult in the non–toilet-trained child, where obtaining a 24-hour urine collection would require an indwelling catheter and bag collection. Spot urine tests for causative factors have been performed and can be helpful in determining specific metabolic abnormalities that the child may have and may help direct preventive treatment.

Decreasing high animal protein intake and sodium intake are important in any dietary recommendations for stone prevention, as is appropriate intake of potassium, calcium, and magnesium, which are protective against oxalate composite stone formation.

Also, potassium citrate (2-4 mEq/kg/d) is a mainstay in preventive treatment in children at risk for stone formation because of underlying metabolic abnormalities, but it is not very palatable and may be hard to give to children for the 2 to 3 times a day that may be needed to improve urinary citrate concentrations. Potassium citrate is found in several clear lemon-lime soft drinks (such as Sprite, 7UP or Mist Twst [formerly Sierra Mist]) that are generally more palatable to children, may be less costly than prescription potassium citrate, are available in sugar-free forms, and have the additional advantage of increasing fluid intake while improving urinary citrate levels.

Radiologic evaluation for urolithiasis in children is predicated on ALARA (as low as reasonably achievable) techniques. Adult patients are generally evaluated with high-radiation testing such as computer tomography for diagnosis, treatment, and surveillance. These high-radiation techniques are rarely needed in children unless there is some uncertainty as to diagnosis or conflicting findings on low-radiation techniques that are generally used.

It is easy to obtain an abdominal flat plate (KUB [kidneys, ureter, bladder]) and ultrasound imaging of the kidneys and bladder, as these tests will...
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find most stones because the majority of stones in children are radiopaque. Furthermore, most large stones in the kidney or ureter near the bladder that may need interventional treatment will be found on these studies. Smaller stones, as mentioned, tend to pass spontaneously and may be inferred if not directly seen by the presence of hydronephrosis or hydro-ureteronephrosis in ultrasonography (Figures 1 and 2).

Surgical techniques
Although many stones will pass spontaneously, some children may require surgical treatment for their stones. Surgical techniques used in removing stones in adults are similarly successful in children. Children should be referred to a pediatric urologist or general urologist if they have stones that are generally larger than 7 mm to 8 mm, as these are less like to pass spontaneously; if their stones do not appear to pass in a reasonable time frame (generally should pass or move from the initial stone location on evaluation in about 4–6 weeks); or if the patient has a congenital or acquired anatomic abnormality increasing the risk of stone formation or decreasing the likelihood of passing the stones spontaneously.

Treatment options include ureteroscopy with or without lithotripsy, percutaneous nephrolithotomy with or without lithotripsy, and extracorporeal shock wave lithotripsy (ESWL). Open and minimally invasive procedures also can be considered but are currently less often used in both children and adults.

Interestingly, although ESWL is less successful when used in adults because of poor stone-free rates, ESWL treatment for renal stones in children, including fairly large stones, tends to be more successful when used in children and achieves reasonable stone-free rates (Figures 3 and 4).

The National Institute of Diabetes and Digestive and Kidney Diseases (NIDDK) has a helpful information publication in English that explains kidney stone formation, management, and treatment that is available online for families and pediatrician offices at www.kidney.niddk.nih.gov (“Kidney Stones in Children,” NIH Publication No. 11-7383, September 2011).

Summary
Urolithiasis is becoming much more prevalent in children of all ages, and it needs to be treated aggressively with prevention. Many of these children will be seen by their primary care physician or nurse practitioner who needs to consider urolithiasis as a diagnosis in children presenting with gross hematuria as well as various degrees of abdominal or colicky pain.

Prevention of stone recurrence depends upon ongoing increased hydration, limited salt intake, and improved dietary intake. Spot urine or 24-hour urine collection for stone risk will help determine certain dietary changes specific for metabolic abnormalities found on evaluation.

Radiographic testing can diagnose stones in most children; however, limiting use of ionizing radiation is imperative to reduce lifetime risk of radiation exposure in children who are likely to have recurrence rates similar to adults.

Most children will not need surgical intervention for their stone disease, but referral to pediatric urology may be necessary for large stones, complicated patient anatomy, and for stones that do not pass in a reasonable time period. The techniques used in adults work equally well in children.

REFERENCES
Small-for-age toddler is unable to walk

Amy Lindley, MD
Shannon G Farmakis, MD
Aline T Tanios, MD

The Case

A 22-month-old African American boy born at 38 weeks by normal vaginal delivery presents to a local hospital from a private pediatric office for failure to thrive. He was seen by his pediatrician until aged 1 month but was lost to follow-up. His delay in walking prompted his mother to reestablish care at age 22 months.

History of illness

There were no reported complications during pregnancy or delivery. According to his mother, the toddler has always been small and has had slow growth, but he has never lost weight. He was exclusively breastfed until aged 18 months, and his current diet consists of 1 cup of whole milk and several cups of fruit juice per day, grains, meats, and minimal fruits and vegetables. He lives with his mother and 3-year-old sister in public housing. His mother has a history of iron deficiency anemia and sickle cell trait. His maternal grandmother has lupus and rheumatoid arthritis. His sister is reportedly healthy and has appropriate growth and development.

Physical exam

On admission, the patient is notably small for his age and is below the first percentile for weight, height, and head circumference. He is alert and cooperative, and is babbling during the exam. His anterior

Figure 1 A radiograph of the patient with failure to thrive and inability to walk was taken at presentation.
fontanelle is open and his oropharynx and dentition are normal. No abnormalities are discovered during a cardiopulmonary exam. There are nodules along his rib cage. His abdominal exam is normal and reveals no hepatosplenomegaly or masses. He has bowing deformities of both arms and legs, wide wrists, and mild diffuse hypotonia. Based on a physical exam and history of developmental milestones, he has both gross motor and fine motor delay.

**Laboratory and imaging tests**

The patient’s blood chemistry panel is presented in Table 1. His urinalysis has 1+ protein (reference range [RR], negative) and is otherwise normal.

A radiograph is obtained at presentation (Figure 1). Because of the findings on imaging, further testing is performed with results as follows: 25-hydroxyvitamin D (25[OH]D), 13 ng/mL (RR, 30-100 ng/mL); phosphorus, 2.99 mg/dL (RR, 4.37-6.59 mg/dL); magnesium, 3.1 mg/dL (RR, 1.7-2.3 mg/dL); and parathyroid, 88 pg/mL (RR, 14-72 pg/mL).

**Differential diagnosis**

The diagnostic considerations of a toddler with failure to thrive are vast (Table 2), but these can be organized into 1 of 3 categories: inadequate intake, malabsorption, or increased metabolic demand. In developing countries, infectious diseases and inadequate nutrition are typically seen as causes for failure to thrive, while in developed countries, causes typically are preterm birth and family dysfunction. Inadequate intake is a possible cause for failure to thrive in this patient as there is significant risk for food insecurity and lack of education regarding dietary needs.

This patient has no evidence of oromotor dysfunction or history of emesis that would suggest these as causes of inadequate intake. There is no history of diarrhea, bloody or fatty stools, or other systemic findings to suggest cystic fibrosis, celiac disease, food protein insensitivity, or other processes causing malabsorption. Causes of increased metabolic demand include congenital infections (ie, human immunodeficiency virus [HIV]), chronic renal disease, renal tubular acidosis, cardiac disease, and vitamin deficiencies. A detailed patient history and review of systems do not suggest any underlying cardiac or kidney disease, but concerns for vitamin deficiency arise.

For most patients with failure to thrive, there is evidence of an etiology found in the patient’s history and physical exam. For 18 months, this patient was solely breastfed without any vitamin D supplementation, and when he transitioned to cow’s milk, the amount he received did not meet daily vitamin D requirements. This, along with the physical exam and radiographic

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**Table 1**

<table>
<thead>
<tr>
<th>TEST</th>
<th>RESULTS</th>
<th>REFERENCE RANGE</th>
</tr>
</thead>
<tbody>
<tr>
<td>Glucose</td>
<td>101 mg/dL</td>
<td>70-105 mg/dL</td>
</tr>
<tr>
<td>Sodium</td>
<td>136 mmol/L</td>
<td>136-145 mmol/L</td>
</tr>
<tr>
<td>Potassium</td>
<td>4.1 mmol/L</td>
<td>3.5-5.1 mmol/L</td>
</tr>
<tr>
<td>Chloride</td>
<td>108 mmol/L</td>
<td>98-107 mmol/L</td>
</tr>
<tr>
<td>Bicarbonate</td>
<td>21 mmol/L</td>
<td>20-28 mmol/L</td>
</tr>
<tr>
<td>Calcium</td>
<td>9.95 mg/dL</td>
<td>9.16-10.96 mg/dL</td>
</tr>
<tr>
<td>Alkaline phosphatase</td>
<td>2139 U/L</td>
<td>150-420 U/L</td>
</tr>
<tr>
<td>Alanine transaminase</td>
<td>9 U/L</td>
<td>6-46 U/L</td>
</tr>
<tr>
<td>Aspartate transaminase</td>
<td>34 U/L</td>
<td>20-65 U/L</td>
</tr>
<tr>
<td>Total protein</td>
<td>6.5 gm/dL</td>
<td>6.1-8.3 gm/dL</td>
</tr>
<tr>
<td>Albumin</td>
<td>4.2 gm/dL</td>
<td>3.0-4.6 gm/dL</td>
</tr>
<tr>
<td>Total bilirubin</td>
<td>0.6 mg/dL</td>
<td>0.3-1.2 mg/dL</td>
</tr>
<tr>
<td>Hemoglobin</td>
<td>10.9 gm/dL</td>
<td>10.5-13.5 gm/dL</td>
</tr>
<tr>
<td>Mean corpuscular volume</td>
<td>68.4 fl</td>
<td>70-86 fl</td>
</tr>
<tr>
<td>Iron level</td>
<td>64 ug/dL</td>
<td>31-144 ug/dL</td>
</tr>
<tr>
<td>Transferrin</td>
<td>331 mg/dL</td>
<td>189-388 mg/dL</td>
</tr>
<tr>
<td>Ferritin</td>
<td>&lt;10 ng/mL</td>
<td>10-140 ng/mL</td>
</tr>
</tbody>
</table>
findings of muscular weakness, fractures, rachitic rosary, enlargement of wrists and ankles, and anterior bowing of long bones are consistent with vitamin D-deficient rickets.

**Discussion**

The 2 main sources of vitamin D are sunlight and diet. Both sources begin as cholecalciferol (D3), which then requires 2 separate steps to become the active form, 1,25 dihydroxycholecalciferol, or calcitriol. Hydroxylation occurs in the liver followed by the kidneys where the final step to create the active form occurs. Regulation by parathyroid hormone and serum calcium levels plays a role in activation of calcitriol. Once activated, 1,25 dihydroxycholecalciferol can now bind to the vitamin D receptor. The calcium-binding protein is formed promoting calcium absorption from the gut, which allows for bone mineralization.3,4,5 The American Academy of Pediatrics (AAP) recommends exclusive breastfeeding for the first 6 months of life and supplementation with 400 IU daily of vitamin D. The recommendation for children aged 1 to 18 years is 600 IU per day. Therefore, if transitioning to cow’s milk around 1 year of age, children should receive 2 to 3 servings of milk per day (16-24 oz/d) to meet this requirement.

Rickets is a result of poor bone mineralization. Without vitamin D, calcium and phosphorous are poorly absorbed, leading to insufficient mineralization.6 These hormonal regulatory factors promote hypertrophic growth plate apoptosis, which allows for the replacement of matrix and eventually leads to bone generation. The hypertrophic chondrocytes are not resorbed because of defective apoptosis and the irregularly calcified growth plate expands.7 This expansion of hypertrophic cells gives rise to the clinical features of rickets: hypertrophy and widening of costochondral junctions, wrists, and knees, and poor mineralization leading to bowed weight-bearing limbs and delayed growth.8,9

The radiologic images of the patient highlight these findings (Figures 2 and 3).

**Typical presentation of rickets**

Although vitamin D deficiency is likely on the rise in the United States, most children with this deficiency do not present with rickets as highlighted in this case, and especially not to this severity. However, understanding the typical presentation helps to identify and treat the disease at an earlier point. Multiple retrospective analyses have shown that the clinical presentation will vary depending on the age of presentation, which is likely related to the skeletal development and motor development in childhood because rickets is typically seen during periods of peak growth.5

A study performed in Spain by Yeste and colleagues10 found that nearly 50% of children with rickets aged from 6 to 12 months present with failure to thrive, and 69% of children aged 12 months and older present with growth failure, seizures, fractures, and laboratory evidence of rickets. Only about 29% of the infants aged 6 to 12 months in the study showed external signs or bony deformities.

A retrospective analysis from

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**TABLE 2**

<table>
<thead>
<tr>
<th>DIFFERENTIAL DIAGNOSIS OF FAILURE TO THRIVE</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Inadequate intake</strong></td>
</tr>
<tr>
<td>• Not enough food given to child</td>
</tr>
<tr>
<td>• Food insecurity</td>
</tr>
<tr>
<td>• Excessive juice</td>
</tr>
<tr>
<td>• Problem with breastfeeding</td>
</tr>
<tr>
<td>• Poor education on dietary needs</td>
</tr>
<tr>
<td>• Not enough food taken by child</td>
</tr>
<tr>
<td>• Oromotor dysfunction</td>
</tr>
<tr>
<td>• Developmental delay</td>
</tr>
<tr>
<td>• Aversion</td>
</tr>
<tr>
<td>• Anorexia</td>
</tr>
<tr>
<td>• Child with emesis</td>
</tr>
<tr>
<td>• Pyloric stenosis</td>
</tr>
<tr>
<td>• Gastroesophageal reflux</td>
</tr>
<tr>
<td>• Malrotation with intermittent volvulus</td>
</tr>
<tr>
<td>• Increased intracranial pressure</td>
</tr>
<tr>
<td>• Rumination</td>
</tr>
<tr>
<td>• Cyclic vomiting</td>
</tr>
<tr>
<td><strong>Inadequate absorption</strong></td>
</tr>
<tr>
<td>• Cystic fibrosis</td>
</tr>
<tr>
<td>• Celiac disease</td>
</tr>
<tr>
<td>• Inflammatory bowel disease</td>
</tr>
<tr>
<td><strong>Increased metabolic demand</strong></td>
</tr>
<tr>
<td>• Congenital infections</td>
</tr>
<tr>
<td>• Malignancy</td>
</tr>
<tr>
<td>• Food protein allergy, insensitivity, or intolerance</td>
</tr>
<tr>
<td>• Short gut syndrome</td>
</tr>
<tr>
<td>• Cardiac, pulmonary, endocrine, and renal disease</td>
</tr>
<tr>
<td>• Metabolic disorders</td>
</tr>
</tbody>
</table>

From Jaffe A; McLean HS, et al.2
Torun and colleagues\textsuperscript{11} also divided subjects into age groups to assess for patterns of symptoms at diagnosis. Children aged from 1 to 3 years present with muscular weakness (91%), failure to thrive (89%), and bony deformities (29%).

DeLucia and associates\textsuperscript{12} assessed the clinical presentation and severity of growth delay by comparing growth parameters. The majority of subjects presented after age 12 months and were breastfed for an average of 12.5 months. Only 20% received appropriate vitamin D supplementation. Skeletal abnormalities and failure to thrive were the 2 most common clinical presentations. Growth parameters of these children were striking, with 65% below the 5th percentile for height and 43% below the 5th percentile for weight.\textsuperscript{12} Extraskeletal findings of patients with rickets include “hypocalcemia leading to tetany, seizures, laryngospasm, and hypocalcemic cardiomyopathy and death.”\textsuperscript{6} Other findings include irritability and delayed motor milestones. Based on these data, the skeletal findings characteristic of rickets are unlikely to be the presenting complaint. Therefore, recognizing vitamin D deficiency as a cause of failure to thrive may prevent severe cases in at-risk populations.

**Risk factors**

Because melanin acts as a barrier to vitamin D synthesis, nutritional rickets is more likely to be found among members of certain racial or ethnic groups with darker skin complexion, such as African Americans, Hispanics, and Middle Easterners.\textsuperscript{10,12-14} Rickets is also more common among infants who are solely breastfed without adequate supplementation or who are poorly transitioned from cow’s milk. It has been shown that breastfed infants of mothers with adequate vitamin D stores will be vitamin D deficient within 8 weeks.\textsuperscript{5}

Rickets is also more likely to develop during winter months at higher altitudes because there is “an increase in the sun’s zenith angle [that] results in an increased path length for the [ultraviolet]B photons to travel,” therefore, little-to-no
vitamin D is produced.5,6 Many of these risk factors likely contribute to the nutritional rickets seen in this patient. Recognizing the clinical and laboratory patterns of nutritional rickets remains essential in the diagnostic process. Acknowledging patients’ risk factors might also serve as a preventive measure to avoid the severity seen in this case.

In general, for children aged from 1 to 18 years who are vitamin D deficient, the Endocrine Society recommends 2000 IU daily of vitamin D2 or D3, or 50,000 IU of vitamin D2 or D3 weekly for at least 6 weeks with a goal serum 25(OH)D level greater than 30.15 It has been shown that in a short-term period with rachitic patients, similar increases in 25(OH)D levels are seen with either D2 or D3. Because it is possible that D2 is metabolized more quickly than D3, many clinicians use D3 for therapy.16 Depending on age, maintenance therapy is 400 IU/d to 1000 IU/d for children aged 0 to 1 year and 600 IU/d to 1000 IU/d for those aged 1 to 18 years.

Follow-up radiologic imaging can be a helpful determinant of recovery. The first sign of recovery is the “healing line of rickets, which is a radio-opaque line in the epiphysis signifying that mineralization of the provisional zone of calcification has begun.”17 Neuromuscular findings, including hypotonia that can lead to motor delays such as those seen in this patient, are well described as features of rickets and are shown to occasionally be reversible with vitamin D repletion and supplementation.18 Given the significant motor delay in this patient, ensuring adequate follow-up and referrals for therapies are other key components in treatment of nutritional rickets.

During follow-up 4 weeks later, imaging shows healing rickets (Figure 4). The patient is transitioned to 800 IU daily with plans to follow every month with serial exams and radiologic imaging.

**Conclusion**

Nutritional rickets, although seemingly rare, continues to be reported within the United States. When seeing patients with growth failure, especially infants and toddlers, one should obtain a detailed dietary history and consider vitamin D deficiency as a possible cause. Most children with failure to thrive do not have an underlying medical cause. However, those that do, such as the patient in this case, will usually have physical exam findings that hint toward a diagnosis. Prompt diagnosis and treatment of patients at risk for nutritional rickets will improve developmental outcomes.

**Patient outcome**

The patient is promptly treated with a 1-time dose of 50,000 IU of D3 during admission followed by 2000 IU of D3 daily until healing is noted on follow-up imaging. He is also given calcium supplementation to avoid hypocalcemia that occurs with rapid remineralization of the bony matrix. The remainder of admission involves a strict calorie count, close monitoring of weight gain, lab surveillance, dietary education, and coordination with social work to ensure the safety of the child and coordination for follow-up.

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For references, go to ContemporaryPediatrics.com/puzzler-0117
Kid care on the slopes

Recognizing first that children have anatomic and physiologic differences from adults helps to provide optimal care for kids in winter sports emergencies.

JEFFREY BROWN, MD, MPH, CPE, FAAP
LESLIE E FISHMAN, MD, MBA, FAAP

Skiing is an enormously popular winter sport for children, teenagers, and families both in the United States and internationally. Estimates put the number of skiers globally at more than 200 million, with children accounting for 13% to 27% of these skiers. According to the National Ski Areas Association, an estimated 10 million skiers and snowboarders made 53.6 million daily visits to US ski areas during the 2014-2015 US ski season. According to a national survey, there are 471 ski areas currently operating in the United States.

The authors are 2 pediatricians who have practiced over the last decade in Eagle County, Colorado, home of both Vail Mountain and Beaver Creek, 2 of the most visited ski areas in the United States. Approximately 20% of all the annual ski visits in the United States occur in Colorado, and Vail is the busiest ski area of the many in the state. Thus, the authors have considerable experience with pediatric skiing-related injuries and emergencies in their pediatric hospitalist and general pediatrics practices.

Injury type and toll

At the authors’ local Vail Valley Medical Center (VVMC), hospital billing data from the VVMC Emergency Department (ED) during the 2014-2015 ski season show there were nearly 4800 patient encounters with a diagnostic accident code E003.2 (activities involving snow; skiing, boarding, sledding). Twenty percent of these VVMC ED visits (nearly 1000) were for children.

78,538
NUMBER OF PEDIATRIC HEAD INJURIES REPORTED TO US EMERGENCY DEPARTMENTS FOR SNOWBOARDING OR SKIING FROM 1996 TO 2010.

and teenagers. Of the 4800 patients who visited the ED, approximately 500 were admitted to the hospital, and 15% of these admissions (about 75 total admissions) were for kids injured while skiing and snowboarding. Fifty percent of these admissions were for unspecified orthopedic injuries and 12% were for a traumatic brain injury.

Clearly, skiing is a sport with a significant risk of injury and, rarely but tragically, death. During the 2012-2013 Colorado ski season, there were 25 skiing-related deaths. Twenty-three of the fatalities occurred while skiing and 2 while snowboarding. In general, there is a consistent 4:1, male-to-female ratio in skier deaths. According to the Colorado Department of Public Health and Environment, 88% of Colorado skier deaths occurred on the slopes, 11% in terrain parks, and 4% were chair-lift associated. Perhaps surprisingly, despite ski helmets’ critical role in injury prevention, about 60% of deaths were sustained by skiers who were wearing a ski helmet.

Epidemiologists estimate that 600,000 people are injured annually in the United States as a result of skiing and snowboarding. Whereas some of these injuries are treated without pursuing medical care, many of the injured will seek care in an ED, acute care facility, or pediatrician’s or primary care provider’s office.

Ski vs board trauma
Skiers are prone to sustain lacera-
tions, boot-top contusions, thumb injuries, and complex knee injuries. In contrast, snowboarders tend to experience distal radius fractures and foot and ankle injuries. Skiers are typically prone to severe injuries sustained from collisions on the slopes, while snowboarders tend to suffer injuries from falls and jumps, not uncommon in terrain parks. Snowboarders are 6 times more likely than skiers to sustain a splenic injury from abdominal trauma (so-called “boarder belly”), with males 21 times more likely to sustain this injury than females.5

In a recent study published by colleagues at VVMC, snowfall and mechanism of injury were reviewed in 644 ski-related hospitalizations.6 The majority of these injuries occurred when there was less than 1 inch of new snowfall, and snowfall of less than 2 inches was associated with increased injury severity. This corroborates the long-held ski patroller observation that, with low snowfall, the slopes are icier and faster and skiers are at increased risk of all injuries under such conditions—particularly severe injuries. In the VVMC study, collisions were associated with the most severe injuries: renal injuries and severe thoracic injuries. Consequently, the authors recommend caution and a thorough evaluation of patients with any injury sustained in a collision.

Kids are different
When caring for pediatric and adolescent patients with skiing-related injuries, it is important to remember the mantra long articulated by pediatricians: “Kids are not little adults.” In regard to any cold weather injuries, keep in mind that children have a larger surface area and thinner skin for their weight. Therefore, they can have more difficulty maintaining body temperature compared with adults. They are also at increased risk of both dehydration and hypothermia when compared with adult skiers and snowboarders.

The implications of these realities are clear: When caring for a child on the slopes, think warmth and hydration. Provide warmth by getting them inside as soon as possible, covering them with blankets and/or additional outdoor ski clothing, supplying hand warmers, and offering warm beverages. Water is the preferred first fluid to start with in caring for children. Avoid sports drinks because of their glucose and electrolyte content.7

Regarding energy needs, kids have faster metabolisms than adults and consequently deplete their glucose stores more quickly. Therefore,
it is a good idea to quickly get some simple sugars such as chocolates, energy bars, juices, and so on into a child suffering a winter sports-related injury.

More differences to consider

Children typically have much healthier hearts than adults, so, provided they have no underlying congenital or other heart disease, an injured child is not likely to have a cardiac problem such as a myocardial infarction or a dysrhythmia. Thus, when coming upon an incapacitated pediatric skier, one should consider a respiratory etiology and not a cardiac problem. Keep in mind, too, when assessing a child for a ski-related injury, that children have higher respiratory rates than adults. Anatomically, children also have relatively small noses, mouths, and tongues and larger tonsils, narrower glottises, and shorter tracheas compared with adults. As a result, children are more predisposed than adults to obstructive-type respiratory problems.

Further, because children have smaller blood volume for body weight relative to adults, they can be prone to hypovolemia even with a relatively small blood loss. Fortunately, children’s bones, joints, and ligaments are more flexible than adults, which enables better absorption of orthopedic trauma. In winter sports mishaps, this results in fewer severe fractures, but more greenstick fractures. However, because children have open growth plates, one must carefully evaluate for possible growth plate fractures (Salter-Harris fractures), as the ramifications of a fracture through a growth plate are significant and need to be carefully managed. Additionally, although, in general, children are at low risk for pelvic fractures, these should not be discounted or overlooked because the consequences can be serious.

Impact of altitude

Acute mountain sickness (AMS) is the effect on the body of being in a high-altitude environment above 8000 feet. Three-quarters of all skiers experience mild symptoms of AMS over 10,000 feet. Common AMS symptoms include difficulty sleeping, fatigue, headache, nausea or vomiting, tachycardia, and shortness of breath. These symptoms typically peak on the first night of arrival to altitude. In general, children are at less risk for AMS and there is no role for pharmacological AMS prophylaxis for children and adolescents. In a recent study, when skiing at altitude, children experienced AMS only about 35% of the time compared with nearly 80% of adult skiers. Most pediatric

Odds of kids wearing a ski helmet were 9.55 times higher if their parent also wore them.

skiers with AMS symptoms do not seek medical care and are treated in the hotel, condominium, resort, and home with rest, over-the-counter medication (acetaminophen, ibuprofen), and hydration.

**Head injuries are of paramount concern**

One of the chief ski-related injuries of concern affecting children or teenagers is a head or brain injury. Skiers experiencing a fall affecting the head can sustain concussions and other brain injuries, skull fractures, scalp injuries, lacerations, or nose and ear injuries. Any child or adolescent who has sustained a fall impacting the head should be thoroughly evaluated for head or brain injury.

Concussions can present with a wide range of physical, cognitive, emotional, and sleep signs and symptoms (Table 1). The decision by the primary care provider regarding concussion management, including neuroimaging, hospitalization, referral to subspecialist/pediatric neurologist, concussion management clinic, and so on can be difficult and challenging. Therefore, a healthcare professional initially evaluating a child or teenaged skier with a head injury should have a low threshold to seek a higher level of care at a hospital, ED, or acute care clinic/facility for a thorough evaluation of the injury. For a complete discussion of concussions and concussion management, the reader is referred to the American Academy of Pediatrics (AAP) Clinical Report—Sport-Related Concussion in Children and Adolescents.10

If a child or teenager does sustain a concussion, in addition to seeking acute care, it is important to be cognizant of the second impact syndrome (SIS). This syndrome, first identified in the literature in 1973, describes the consequences of a patient sustaining a second blow or concussion before the first concussion has healed and resolved. This second impact, even if less severe than the first injury, can have a devastating outcome with the injured child/teenager falling to the ground, rapidly showing signs and symptoms of increased intracranial pressure. This syndrome has been the cause of death for a number of high school contact sport athletes.

The parents/family of a child or adolescent who sustains a head injury/concussion at a destination resort should be made aware of the risk of SIS and be advised to promptly consult with their pediatrician/primary care provider when home regarding further concussion evaluation, management, and return to school and play. Further, the child or teenager having sustained the head injury should not resume skiing during his or her vacation unless it is clear that the concussion has resolved.

### Helmets for prevention

Ski helmets have become an indispensable part of equipment for kids and adolescents who are skiing and/or snowboarding. In skiing and snowboarding, head injuries account for 9% to 19% of all ski-related pediatric injuries reported by ski patrols.

### Table 1

<table>
<thead>
<tr>
<th><strong>PHYSICAL</strong></th>
<th><strong>COGNITIVE</strong></th>
<th><strong>EMOTIONAL</strong></th>
<th><strong>SLEEP</strong></th>
</tr>
</thead>
<tbody>
<tr>
<td>Headache</td>
<td>Feeling mentally “foggy”</td>
<td>Irritability</td>
<td>Drowsiness</td>
</tr>
<tr>
<td>Nausea</td>
<td>Feeling slowed down</td>
<td>Sadness</td>
<td>Sleeping more than usual</td>
</tr>
<tr>
<td>Vomiting</td>
<td>Difficulty concentrating</td>
<td>More emotional</td>
<td>Sleeping less than usual</td>
</tr>
<tr>
<td>Balance problems</td>
<td>Difficulty remembering</td>
<td>Nervousness</td>
<td>Difficulty falling asleep</td>
</tr>
<tr>
<td>Visual problems</td>
<td>Forgetful of recent information</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Fatigue</td>
<td>Confused about recent events</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Sensitivity to light</td>
<td>Answers questions slowly</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Sensitivity to noise</td>
<td>Repeats questions</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Dazed</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Stunned</td>
<td></td>
<td></td>
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</tbody>
</table>

From Halstead ME, et al.10
In recently published literature including a meta-analysis, ski helmet use was found to reduce head injury risk by as much as 35% and did not increase the risk for a neck injury while skiing or snowboarding.16-18 This evidence-based data has helped to further successful advocacy for ski helmet use for children and teenagers.

At Vail Mountain, resort policy is that all children and teenagers in a ski school lesson or program must wear a ski helmet. Recent observational studies performed by VVMC staff at the resort have found ski helmet use rates overall to be 80% for all participants and consistently over 95% for the kids and adolescents skiing there (according to Kim Greene, VVMC Think First Program). Additional studies have demonstrated that ski helmet wearing has minimal effect on hearing,19 does not diminish vision,20 and, in fact, leads to a higher safety awareness on the slopes.21 Both education and helmet laws have been shown to increase ski helmet use among children and teenagers.22

A final key point to remember is that if a child has a marked fall on the head with a ski helmet, that helmet has done its job and should be replaced by a new helmet.

**The pediatrician’s role**

Pediatricians can be invaluable sources of information, support, and advocacy on the slopes, in the office, and in the community to promote the use of ski helmets for children and adolescents and to be aware of the management of other winter sports risks and injuries.

The authors advocate that pediatricians who see patients and families who ski and snowboard provide anticipatory guidance to parents regarding helmet use for themselves and all skiing or snowboarding members of the family. As with the successful advocacy of bicycle helmet use, the pediatrician can be a welcome source of information and support to help prevent winter sports head and brain injuries by promoting ski helmet use among the patients and families in their practices.

A summary of the important considerations for the pediatrician/medical provider caring for a child or adolescent with a skiing-related injury or emergency is presented in Table 2.

**Conclusion**

Skiing and snowboarding can be immensely fun and enjoyable activities and good exercise for kids, teenagers, and families. However, when caring for a child or adolescent on the slopes, or in the lodge, ski patrol station, acute care setting, or office, it is vital to remember that, as in other areas of pediatrics, kids are not little adults. Remembering their unique anatomical and physiological differences can help the pediatrician provide optimal medical care and advice that can lead to good outcomes in the event of injuries or emergencies sustained on the slopes.

**Table 2**

<table>
<thead>
<tr>
<th>Caring for a Child with a Ski-Related Injury or Emergency</th>
</tr>
</thead>
<tbody>
<tr>
<td>1. Think warmth, hydration, and simple sugars.</td>
</tr>
<tr>
<td>2. Think lungs before heart. For the compromised pediatric skier, think respiratory, not cardiac, etiologies.</td>
</tr>
<tr>
<td>3. Think softer bones and different fractures. Don’t miss greenstick fractures even if there are no compound fractures. SALTER-HARRIS fractures need careful attention in children and adolescents.</td>
</tr>
<tr>
<td>4. Acute mountain sickness (AMS) affects children less than adults, but it should not be overlooked.</td>
</tr>
<tr>
<td>5. Abdominal internal organ injuries can occur and not be obvious. Consider “boarder belly” in snowboarders who have abdominal trauma.</td>
</tr>
<tr>
<td>6. Ask about underlying medical issues: type 1 diabetes, asthma, seizure disorders, and so on.</td>
</tr>
<tr>
<td>7. Carefully consider any possible concussion and keep the second impact syndrome (SIS) in mind.</td>
</tr>
<tr>
<td>8. Think prevention. Promote ski helmet use for children and teenagers who are skiers and snowboarders.</td>
</tr>
</tbody>
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For references, go to ContemporaryPediatrics.com/winter-sports-injuries

*Approximately 15% of downhill skiing injuries among kids and teens are caused by musculoskeletal immaturity.*

Unhealthy prenatal diet linked to ADHD

First-of-its-kind study connects high-fat, high-sugar diet with ADHD in offspring.

CRYSTAL MURCIA, PHD

A longitudinal study examined the relationship between prenatal or postnatal high-fat, high-sugar diet and symptoms of attention-deficit/hyperactivity disorder (ADHD) in children who demonstrated either early-onset persistent conduct disorder or minimal conduct problems.

Conduct disorder and ADHD often coexist, and, when found together, increase the likelihood for behavioral problems, academic underachievement, substance abuse, social maladjustment, and poor self-esteem. Conduct disorders fall into different categories based on their developmental course (early-onset persistent [EOP], childhood limited, and adolescent onset), but are all generally characterized by repeated violation of the rights of others and lack of age-appropriate conformity to rules or societal norms.

Previous studies have linked an unhealthy prenatal maternal diet (ie, consumption of foods that are high in fat and sugar) with ADHD and other neuropsychiatric disorders in their offspring. The putative effect of such a diet is to increase DNA methylation, which in turn influences gene expression. One gene whose methylation is known to incur persistent changes in methylation due to diet is insulin-like growth factor 2 (IGF2). The IGF2 gene also has developmental significance, as it has been linked to placental and fetal growth as well as to postnatal brain development.

Looking for connections

In a first-of-its-kind study, Jolien Rijlaarsdam, PhD, Leiden University Centre for Child and Family Studies, Leiden, The Netherlands, and colleagues evaluated the relationships between a high-fat, high-sugar diet (ie, an unhealthy diet) during the prenatal and postnatal periods, IGF2 DNA methylation, and ADHD symptoms. The results, published in the Journal of Child Psychology and Psychiatry, indicate that there are connections between these factors.

The study population consisted of 164 children from the Avon Longitudinal Study of Parents and Children who were identified as having either EOP or a low conduct-problem trajectory (children with childhood limited or adolescent onset conduct disorders were not included). Prenatal and postnatal diet were determined by parental self-report; IGF2 DNA methylation was quantified using blood samples collected at birth and at age 7 years; and ADHD symptoms were evaluated using a validated instrument at ages 7, 10, and 13 years.

Among the various associations detected, investigators found that children classified as EOP had an overall greater burden of ADHD symptoms compared with low conduct-problem children. In both the EOP and low conduct-problem groups, a prenatal unhealthy diet correlated with a greater extent of IGF2 methylation at birth. In the EOP population (but not the low conduct-problem group), increased IGF2 methylation at birth was associated with a greater ADHD symptom burden. Because of the link between a prenatal unhealthy diet and IGF2 methylation at birth, an unhealthy prenatal diet indirectly correlated with ADHD symptoms in children with EOP.

Notably, IGF2 methylation was not associated with other psychiatric disorders assessed (oppositional defiant disorder, generalized anxiety disorder, and major depressive disorder) in either the EOP or low conduct-problem groups.

The researchers conclude that “Preventing ‘unhealthy diet’ in pregnancy might reduce the risk of ADHD symptoms in EOP youth via lower offspring IGF2 methylation.”

Dr Murcia is a medical writer in North Carolina. She has nothing to disclose in regard to affiliations with or financial interests in any organizations that may have an interest in any part of this article.

For references, go to ContemporaryPediatrics.com/prenatal-diet-and-ADHD
What motivates teens to tan

Pop culture, social interaction—even addiction—may compel adolescents’ tanning bed habits.

LISETTE HILTON

When dermatologist Cindy Firkins Smith, MD, was conducting research on the topic of teenagers and tanning for a presentation at the 2016 42nd Annual Meeting of the Society for Pediatric Dermatology, she says it surprised her that her assumptions about children’s tanning behavior were not always correct. Smith, a clinical professor of dermatology at the University of Minnesota, Minneapolis, is more aware of the issue than most. She and colleagues worked for 11 years on legislation to prevent minors from using tanning booths.

So, what surprised her? “I assumed that everyone tanned because they wanted their skin to be darker. But in doing the research, I discovered that not everyone’s motivations are the same,” Smith says. “In order to encourage people to stop doing this specific behavior, you really have to know what motivates them.”

Why teenagers tan

Smith cites research by JK Robinson, MD, and colleagues published in 2010 in the Archives of Dermatology as being among the studies to suggest that the primary reason adolescents tan is because they think it makes them look better. “Maybe it’s the difference in their skin color; maybe it’s the difference in the way their clothes look on them; maybe it’s that they look thinner. It’s different for everybody,” she says.

Dermatologists who understand what’s motivating their young patients to tan can appeal to patients’ desires and send more effective messages about the alternatives. For example, Smith says that talking about tanning and skin cancer risk isn’t generally something to which kids will respond. They’re more likely to respond to warnings about how the tan will make them look—what the ultraviolet (UV) light exposure is doing to increase pigmentation and wrinkles that will start to become etched on their faces in their early 20s, she says.

So, offer alternatives that satisfy teenagers’ desire to look better. Some options: sunless tanners, a change in makeup or hairstyle, or a change of clothing colors that look better against pale skin. “My mantra is, ‘Love the skin you’re in,’” Smith says.

Smith makes it a point to tell pediatric patients of all ages—even toddlers—that their skin color is gorgeous, and she gives them and their parents recommendations about how to keep skin free of damage for the long term.

“I tell them they should do a really good job of keeping that skin gorgeous,” she says. “But, if a teenager is committed to having darker skin, then we talk about sunless tanners. I’ve had pretty good success with some of my really fair kids.”

Pop culture plus

Whereas tanning was all the rage in the 1970s, today’s celebrities often flaunt light, sun-damage-free skin. Some examples are Anne Hathaway, Nicole Kidman, Taylor Swift, and Katy Perry. Dermatologists can give those examples to their patients, Smith says.

There are other reasons that kids say they want to tan. “We learned from [the reality television show] Jersey Shore that sometimes tanning is considered a group activity. People might do it for social interaction. If that’s the motivation, suggest better alternatives that don’t involve damaging their skin,” Smith says.
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“Some people do it to relax. We’ll point out that this really isn’t a good way to relax because of the damage it does to your skin, [and ask] ‘Have you considered trying yoga?’ ‘Meditation?’”

There’s still other data that UV exposure is an addictive behavior, stimulating areas of the brain that are stimulated by narcotics. “So, there are people who may think they can’t give it up. And I try to find out why they can’t and address that issue,” she says.

Compliment and counsel

It might take a minute during a clinical appointment to compliment a young patient on her skin and way she looks. Physicians should never miss the opportunity, regardless of the reason for the patient’s visit, Smith stresses. “I want them to embrace who they are. And what they are. They’re beautiful in the skin they’re in,” she says.

It’s important to address teenagers’ tanning motives, Smith points out, because, despite public awareness campaigns and even legislation that is now in some 13 states banning adolescents aged younger than 18 years from accessing tanning booths, the desire to tan remains stronger than the threat of skin cancer.

One of the things that drives Smith is the memory of a young female patient she lost to melanoma. “She was diagnosed with melanoma in her 30s. She was a big tanning booth user. Before she died, she asked me to do anything I could to prevent the same thing from happening to others,” Smith recalls. That patient’s story has resonated with legislators and young patients in her practice, she says.

Other messages that seem to connect and and make a difference, in Smith’s experience, include talking to young girls about how the color of a prom dress might be just the right color to show off their beautiful skin. The dermatologist says that in her 26 years of practice, she’s had scores of kids come back and say how a recommendation she made about how to complement their skin made them realize they didn’t need a tan.

“Sometimes, as physicians, we think we’re powerless, but we’re really not,” she says.

Ms Hilton is a medical writer who has covered health and medicine for 25 years. She resides in Boca Raton, Florida. She has nothing to disclose in regard to affiliations with or financial interests in any organizations that may have an interest in any part of this article.

READ MORE ON THIS TOPIC

Sun protection steps parents take for themselves and their perception of skin cancer risk both influence kids’ sun protection behavior. Therefore, sun protection for children needs to be aimed at parents as well as children. Communicating with parents in a way that incorporates the principles of motivational interviewing may be more effective in promoting behavioral change than admonitions to use sunscreen.

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CONTINUED FROM PAGE 7
making its repeal and replacement a top priority, and Verma will play a significant role.

It’s unusual for a CMS head to have Medicaid but not Medicare experience, says Joan Alker, director of the Georgetown University Center for Children and Families, Washington, DC.

Alker says she is concerned about the vision within Verma’s Medicaid work in several states. Proposals involved more barriers to coverage, such as work requirements, requirements to pay, or disenrollment for missing a deadline for renewal. The Obama administration stopped some of those plans, says Alker.

In Indiana, the provisions impacted adults, but, says Alker, “We do know that as parents are covered, children are more likely to be covered.” The bigger issue, she says, are potential threats to children’s coverage, including the proposed repeal of the Affordable Care Act; the fact that the Children’s Health Insurance Program (CHIP) needs to be re-funded by Congress next year; and potential significant cuts to Medicaid.

In the meantime, on December 15 the Medicaid and CHIP Payment and Access Commission (MACPAC), a congressional advisory panel, called for 5 more years of CHIP funding as soon as possible, both to continue the coverage of children under it and to mitigate states’ budget uncertainty.

**Time for dialog on vaccines**

Will the Trump administration promote vaccine hesitancy or create policies that are less than vaccine friendly? Or will his doubts about vaccines encourage hesitancy?

The vaccine community certainly needs to begin relating to the administration “in a positive dialog,” says Kathryn M. Edwards, MD, FAAP, co-author of the September 2016 American Academy of Pediatrics (AAP) clinical report “Countering vaccine hesitancy.”

Speaking of the president-elect, Edwards says, “Certainly someone who understands businesses and understands numbers should understand that there has probably never been anything more impactful in child health—other than clean water—than vaccines.”

Preventing children from getting disease by vaccinating is not only a smart health decision, but a smart business decision, she says.

Fears about vaccine hesitancy were again stirred when then-candidate Donald Trump met last summer with discredited researcher Andrew Wakefield and other like-minded people. Wakefield engineered much vaccine hesitancy with a fraudulent 1998 study, later retracted, suggesting a vaccine could cause autism.

Trump has repeatedly been quoted as saying immunizations have something to do with the autism “epidemic.”

In the Republican candidates’ debate on September 16, 2016, Trump said, “I am totally in favor of vaccines. But I want smaller doses over a longer period of time. Because you take a baby in—and I’ve seen it—and I’ve seen it, and I had my children taken care of over a long period of time, over a 2- or 3-year period of time.”

He went on to say he knew a recent instance in which a baby “got a tremendous fever, got very, very sick, now is autistic.”

Edwards says she is hopeful the newly named Health and Human Services Secretary, Tom Price, MD, as a physician will understand vaccines’ value. The Republican congressman from Georgia is an orthopedic surgeon and previously chaired the House of Representatives Budget Committee.
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Improve your practice: Facilitate patient access

As patients abandon their medical homes for retail clinics and telehealth visits, let’s remove the obstacles that hinder patients’ access to our services.

Those of us in the trenches of medical care are always seeking new ways to improve our practices. This means we must be willing to try new things, and not infrequently we are often pleasantly surprised when our experiments succeed!

Over the past few years, government bureaucrats have spent a lot of time and effort informing physicians how best to provide medical care. The consequences of this healthcare overhaul have been high-deductible insurance, burned-out physicians, and frustrated patients. While we optimistically wait for healthcare “reform” to be reformed, there is much we providers can do to improve the care we offer patients.

A major dilemma for patients is simply how to access good care that is both convenient and affordable. Complicated electronic health records (EHRs) and paperwork have reduced the number of patients a physician can see per day. Patients often cannot be seen by their primary care provider (PCP) because of long waits on the phone and triage systems that often misdirect patients to emergency departments. Thus, patients are abandoning their “medical home” and seeking care elsewhere. This means that patients are using retail-based clinics that have extended hours and competitive prices for the uninsured or underinsured, and are being tempted to utilize the $49 telehealth visits promoted by some insurance plans. To thrive in these challenging times, practices should consider all options to facilitate patient access. If you have an open mind, you may even consider changing your “traditional” practice to one that provides “direct primary care.” Read on.

Remove obstacles

Before we continue, it’s worth mentioning that there are numerous ways to make pediatric practices more efficient, with the goal of increasing capacity beyond 20 patients a day. Simply by increasing capacity by just a few patients each day, you improve patient access (and increase practice revenue).

Methods that can help accomplish this goal include: 1) improving use of EHRs (or using scribes); 2) using technologies to expedite diagnosis and screenings; 3) having age-appropriate scales in exam rooms (to improve traffic flow); and 4) having staff assume new responsibilities (recording chief complaints, giving vaccines if they do not do so already, and so on) so that the provider’s menial chores are minimized when you enter exam rooms.

Adding capacity also means that you have a mechanism is place to optimize scheduling of patients. This means minimizing “no-show”
appointments by enforcing a strict office policy that makes these events rare. It also means anticipating the need for sick visits by keeping same-day slots available depending on the day of the week and the season of the year. Most practices see more patients on Mondays and Fridays, with fewer patients seen during the middle of the week.

You can also improve patient flow by adopting a “wave method” of scheduling appointments so that you book 2 patients for the same time slot and see whoever shows first. Many practices that use this system report that it improves workflow significantly. Another option is to provide evening hours if you anticipate that you will see more patients with extended hours and not merely displace patients that are usually seen during the day to your evening hours.

I’ve spoken to some pediatricians who have lost patients to convenient care clinics in their neighborhoods. They have regained patient volume and allegiance by implementing a walk-in clinic type of practice during certain hours, and staffing appropriately. The bottom line is, if you are willing to innovate, you can improve upon your present system and increase capacity without working harder. You can even consider adopting a very successful system for scheduling patients that is nearly 20 years old, called Open Access Scheduling (OAS).

Open Access Scheduling
The OAS system was invented by Mark Murray, MD, MPA, and Catherine Tantau, BSN, MPA, at Kaiser Permanente in Northern California in the early 1990s. The essence of this system is scheduling patients on the same day they call, no matter what type of visit is requested. So, rather than having wait times in the order of 50 days to see a patient’s PCP, OAS changes this to a system that facilitates same day visits with PCPs!

Under the traditional model, a provider may be fully booked on any day, and if no same-day slots are kept open—and overflow patients must be seen—the provider becomes double booked. This overwhelms providers and staff. The alternative model is a carve-out system in which at least 50% of visits are booked ahead of time, with the remaining number of slots kept open dependent on the day of the week and season, as well as the capacity and work habits of the PCP.

Under the OAS model, the number of prebooked visits falls to around 30%, and these represent recently booked patients who prefer not to be seen on the day they call. To make OAS work, providers need to clear up any “backlog” of visits, most notably preventive health visits, which can take some practices weeks or months depending on the willingness of providers to pitch in and work extra hours. Practices that wish to implement an OAS system can consider adopting scheduling portals such as Appointment Quest (www.appointmentquest.com/) that enables patients to book their own appointments without calling.

Practices that have implemented the OAS model, once fine-tuned, rave about how it expedites care and pleases patients.
and secondly if they would like to come in for a visit that day. According to Mark Murray, in the open access system “providers do today’s work today,” rather than chipping away at a backlog of work. This model is particularly relevant now, when patients are seeking care at retail-based clinics because they can’t get in to see their own physicians.

**Panel size matters**

If you wish to consider adopting the OAS model discussed above or just want to make your present system work better, you need to realize that physicians have a limit as to the number of patients they can accommodate in their panels. To function as a PCP, one must attend to all the needs of patients, not just see patients for preventive health and ill visits. This involves calling patients to address concerns, generating referrals and school forms, refilling medications, and more.

Panel sizes have been extensively analyzed by experts, who have developed complicated algorithms for computing ideal panel sizes. Long story short, however, a full-time pediatrician’s panel typically caps out at 1500 to 2000 patients, depending on the complexity of patients, number of daily available appointments, and number of support staff. It is unfair to the patients to bloat panels beyond that which the practice can handle. Once a provider panel gets to a critical size, it should be closed and patients directed to other providers in the panel, or additional providers should be hired.

**Direct Primary Care**

The Affordable Care Act (ACA) has led some physicians to create a new model of healthcare delivery, one that can bypass insurance coverage and enable physicians to focus on medical care. This model is called Direct Primary Care (DPC). It entails having patients subscribe to your practice for a monthly or yearly fee. No insurance companies are billed and no co-pays are collected. Patients are advised to have catastrophic health plans in case they need hospitalization, medical tests, or surgery.

In the DPC model, physician panels are capped at 600 to 1000 patients, allowing physicians to do well economically while affording patients ready access to their PCP. In most DPC models, office testing is billed to the patient at cost. Doing the math indicates that if you have patients pay $60 per month per patient ($720) per year, a 1000 patient panel generates $720,000 per physician per year. Even if your overhead remains unchanged, then physicians often make more than they would under traditional models of care. Usually the overhead decreases as well because you are no longer billing patients, and you hire fewer staff because you see fewer patients per day than you would under the traditional care model.

Physicians as well as patients praise this model because wait times are minimal, and visits are longer and unrushed. Obviously, you could not transition to a DPC model if you have a significant Medicaid population in your practice.

**Food for thought**

There are a multitude of ways physicians can facilitate patient access to care. Choices range from improving practice workflow so more patients can be seen per day, to making changes to your scheduling system. Physicians who wish to change their practice model entirely might consider adopting a DPC model of care. Although the choices are many, only you can decide if your current system is working well. If not, consider the many options discussed in this article.

**Send your comments to catherine.radwan@ubm.com**

**Direction Primary Care entails having patients subscribe to your practice for a monthly or yearly fee. No insurance companies are billed and no co-pays are collected.**
Etiology
Mastocytosis is a condition characterized by the accumulation of mast cells and CD34+ mast cell precursors in the skin or other organs of the body. The cause of mastocytosis is unknown but it has been associated with a point mutation (D816V) in the c-KIT stem cell ligand on mast cells, leading to unregulated stimulation of mast cell receptors and mast cell degranulation.1 This mutation may result in release of mast cell mediators including histamine, prostaglandin D2, heparin, tryptase, chymase, leukotrienes, and others.

Potential triggers of mastocytosis can be physical and environmental, as well as reactions to medications and certain foods (Table).

Mastocytosis may occur in individuals from birth to middle age, but most commonly occurs before the age of 2 years.2 In pediatric patients, mastocytosis typically has an early onset before age 1 year and improves without treatment. Up to 25% of pediatric cases may be congenital. Mastocytosis in pediatric patients rarely infiltrates internal organs.

Clinical manifestations
Cutaneous manifestations include solitary mastocytomas, urticaria pigmentosa (UP), diffuse cutaneous mastocytosis, and telangiectasia macularis eruptive perstans.2,3 Blistering and vesiculation may occur because of histamine-mediated or other chemically-mediated leakage resulting in detachment of the epidermis from the underlying dermis. Patients often present with a positive Darier sign, representing localized erythema and urticarial wheals after rubbing the papule.

Rarely, patients may develop systemic symptoms including flushing, blistering, itching, hives, nausea, abdominal pain, diarrhea, bone pain, hypotension, or anaphylaxis.2 The risk of anaphylaxis is comparable to the general population in patients with solitary mastocytomas or UP. It is seen in 9% of pediatric patients (slightly increased from the general population) with severe cutaneous manifestations.3 Mastocytomas are solitary fixed, flesh-colored to hyperpigmented papules or plaques often with a peau d’orange (orange peel-like) surface. Local edema may be severe enough to cause blistering, particularly in infants. These lesions are more often located on the neck, upper extremities, or trunk.

Urticaria pigmentosa appears as 2 or more well-demarcated, reddish-brown or golden-brown macules and papules located on any cutaneous surface, including mucous membranes, but often sparing the palms and soles. It is the most common presentation of mastocytosis with improvement of lesions similar to the course of solitary lesions. These patients may be more prone to the development of itching and subtle systemic symptoms when multiple mastocytomas are irritated.

Diffuse cutaneous mastocytosis features diffuse thickened, erythematous reddish-brown to yellow plaques with a peau d’orange surface. This condition is rare and patients are more likely to have associated systemic manifestations,
including gastrointestinal symptoms such as abdominal pain, nausea, vomiting, diarrhea, or gastrointestinal hemorrhage secondary to gastritis or peptic ulcer disease. Infants and young children are also at risk for mastocytosis syndrome, which includes bronchospasm, headache, flushing, diarrhea, pruritus, hypotension, and occasionally death.

Telangiectasia macularis eruptive perstans manifests as small, red-brown, telangiectatic macules on the trunk or extremities, with little tendency toward urtication. This condition is extremely rare and most patients are not symptomatic.

Differential diagnosis
The differential for brown macules, papules, and plaques includes pigmented nevi, postinflammatory hyperpigmentation, lentigines, and café au lait macules. The golden-brown color and fuzzy borders as well as Darier sign help to distinguish mastocytomas from these pigmented lesions. The differential for bullous lesions includes bullous impetigo, herpes simplex infection, epidermolysis bullosa, contact dermatitis, or linear immunoglobulin A dermatosis. Patients with cutaneous and systemic manifestations may be misdiagnosed with carcinoid syndrome or amyloidosis.

Diagnosis of UP is primarily clinical with a positive Darier sign and occasionally positive dermatoglyphism. Laboratory studies and skin biopsy are not routinely required.

Management
Most patients with mastocytosis are asymptomatic and their condition improves by the time they reach puberty.²,⁵ Symptomatic patients may respond to antihistamines (eg, hydroxyzine, diphenhydramine, cyproheptadine, chlorpheniramine) daily, and they should avoid known triggers (such as hot baths or extreme exercise that may increase histamine release). Other possible treatments include doxepin, leukotriene antagonists, or cromolyn sodium (mast cell stabilizer). However, these treatments are usually reserved for patients with severe cutaneous symptoms or systemic manifestations. Photochemotherapy (PUVA) is rarely used to treat diffuse infiltrative mastocytosis or improve the cosmetic appearance of the rash.

Of note, practitioners should be aware of anesthesia-related adverse events associated in pediatric patients with mastocytosis.⁶ A baseline serum tryptase may be helpful in identifying systemic mastocytosis. Practitioners should avoid preoperative drug skin testing and minimize mechanical cutaneous pressure. Nonsteroidal anti-inflammatory drugs may be given with caution to help with flushing, but may also result in further degranulation.

Outcome
The patient was growing and developing normally and did not require any treatment.

Dr Goel is a second-year pediatric resident at Rainbow Babies and Children’s Hospital, Cleveland, Ohio.
Dr Cohen, section editor for Dermcase, is professor of Pediatrics and Dermatology, Johns Hopkins University School of Medicine, Baltimore, Maryland. The author and section editor have nothing to disclose in regard to affiliations with or financial interests in any organizations that may have an interest in any part of this article. Vignettes are based on real cases that have been modified to focus on key teaching points. Images also may be substituted for teaching purposes.

For references, go to ContemporaryPediatrics.com/dermcase-0117

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Recurrent brown spots in an infant girl

NIKITA GOEL, MD, PGY2

THE CASE

A mother brings her healthy 6-month-old girl to the outpatient clinic with disseminated, asymptomatic, golden-brown bumps that occasionally become red and swollen. For more on this case, turn to page 37.
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