

# PEDIATRIC NEWS

## San Antonio Military Pediatric Center



Wilford Hall Air Force Medical Center  
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### Incorporating Teachable Moments

Laura Place Maj MC  
San Antonio  
Military Pediatric Center

**I**magine a typical day in your practice ....  
The waiting room is full, the patient's chart hits your door 15 minutes after their scheduled appointment time, you quickly try to complete the myriad of forms and questions that a well baby visit entails, and as you are winding up the visit, the inevitable occurs ... "oh, by the way, what do you think about ....." You sigh and try to contain your exasperation. The last thing you want to be doing is starting a conversation about sleep, discipline, or feeding challenges during the final seconds of an appointment.

Despite how and when they bring up these concerns, their child's behavior and development are often the key issues for most parents during a well child visit. Most importantly, they want to know: Is my child doing OK – i.e. is she developing OK? And

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### Tattoos, the Other Pigmented Lesion

Jeffrey Meffert, Col MC  
Program Director, Dermatology  
San Antonio Uniformed Services Health Education Consortium

**T**attoos have always been with us and are certainly part of the current culture. The best advice for teens considering getting a tattoo is the same as that for teens considering sexual activity: "No". This advice, unfortunately, may be ignored on both counts and the pediatrician will now have a tattooed teenage parent for a patient. Like sexual activity, there is a measure of pre-activity counseling which may save a lot of grief, heartache, and expense later on. Years ago when Ann Landers was asked by an anxious parent about their newly tattooed child, she advised a trip to the doctor for quick and painless laser removal. This statement proves Ann Landers did not have a tattoo or, at the very least, never attempted to have hers removed. There are a number of myths about tattoo removal, which should be addressed, in the aforementioned counseling. These myths include that treatments are fast, painless, and complete. Other myths are that everyone has them and this permanent alteration is a good way to express your self. The basic counseling recommendations are:

1. No amateur tattoos and get them in a clean professional setting where needles are new for each client
2. Keep it black
3. Keep it small enough to cut out if need be
4. Keep it in a place easily covered by normal clothing
5. No names
6. No violent, racist, satanic, sexual, or obscene text or images
7. Don't get it while intoxicated (more likely to violate rules 1-6)

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### What Causes Mental Retardation?

Scott D. McLean, M.D.  
FAAP, FACMG  
Chief of Medical Genetics  
San Antonio Military Pediatric Center

**G**rowing up in Baltimore, I had a large and tight-knit group of neighborhood friends, and we had opinions about everything. There was one little kid, Stevie, who was different. My friend Fritz said, "He's mental." To us that meant "insane" or "crazy," but in truth Stevie was mentally retarded, and we had a bit of a hard time with that. He couldn't keep up, though he tried. His six older brothers and sisters kept a close

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What can go wrong? Beyond the continuing social stigma tattoos represent in many sectors of society, infectious diseases acquired during tattooing are a valid concern. The old adage “Two anchors, one chance” addresses the possibility of transmission of blood borne diseases, syphilis in this case, by improperly sterilized instruments. This is especially concerning when getting amateur tattoos in which dirty implements may be reused on multiple clients. While there has never been a documented case of HIV transmission through professionally placed tattoos, the risk of Hepatitis B and Hepatitis C is well documented. It has even been postulated that the intradermal, rather than intravenous, injection of virions may predisposed to sub clinical HCV infection. This risk of infection is why potential blood donors are banned for a time period after their tattoo. Some states have tighter regulation of tattoo parlors than others but in no states are inspections regular. It is the rare state, which has routine health inspections for the artists.

Other inflammatory conditions may occur in relation to a tattoo, most notably a sarcoidal or granulomatous skin reaction. Pseudolymphoma changes have also been identified in chronically inflamed tattoos. The FDA does not regulate tattoo dyes and artists tend to have their own secret recipes for various dye colors. Modern colors are mixes of several different pigments and a successful, problem free result from one artist or with one set of colors does not guarantee continued luck. We have also seen chronic urticaria precipitated by tattoos, in some cases worsening when attempts were made at laser removal.

More peculiar complications of tattoos include the pigment uptake

by regional lymphatics node mimicking a melanoma positive node on lymph node dissection. A generalized eczematous eruption followed the laceration of a cinnabar-containing tattoo. Permanent cosmetics are tattoos and the metals in the dye can interfere with MRI scans. Even the person considering a tattoo who opts for a more temporary henna tattoo may experience allergic contact dermatitis to the dye. Tattoos, like piercings, should not be done while a patient is on isotretinoin or has recently completed a course of the same. The wounded skin may respond with a pyogenic granuloma-like healing response.

Keep it black. Current laser technology for tattoo removal works best on black (blue-black in situ) pigments. Even in multicolored tattoos, the outlines are generally in a black ink which usually is carbon (India ink) based. Unfortunately melanin absorbs laser energy in this spectrum and a potential risk of laser removal, especially in the patient with his or her own innate pigment, is that normal skin color will be ablated along with the tattoo. In fact, it may be more sensitive to the laser energy and while it will eventually return in most cases, this is not guaranteed. The amateur tattoos are generally black and placed more shallowly than the professional tattoos. These will usually respond more completely to fewer treatments. This is where we run up against two myths: Treatment is fast and treatment will be complete. It may take six to twelve monthly treatments to reach optimized treatment of a professional black tattoo (Amateur tattoos will often achieve maximal benefit with three to six treatments). Even at this time the tattoo may be completely intact, partially smudged, or mostly gone although with some residual skin texture and

pigment changes. In darker skinned individuals, there may be a pale “ghost tattoo” where the darker design once was. In any case, removing a tattoo is much more painful for each of the dozen or more treatment sessions than it was to put it on. Many patients require topical or injectable anesthesia to tolerate the pain.

What if removal must be quick? There are ways to make a tattoo gone quickly but these involve excision (discussed below), dermabrasion, laser vaporization (burns the tattoo away), salabrasion, and aggressive chemical peels. Any of these procedures vigorous enough to remove the tattoo, will leave scars which, whatever the method, usually look like a bad burn. Although this requires a great deal of operator expertise, is painful, and is prone to local infectious complications, it is sometimes the preferable route. Employers will not always judge someone’s personality by their burn scars. Tattoos are another story.

Keep it small enough to cut out. Our current technology is okay at black tattoos. It does poorly with multicolored designs. Reds do not respond well, greens hardly at all, and modern pastels and iridescent colors often will not even budge a little, however many treatments are attempted. Even worse, certain yellows and whites will photo-reduce under the laser and become an ugly mottled gray that does not respond to any current laser therapies. If one is bothered by an inflammatory reaction to the tattoo (sarcoidal or urticarial), applying

#### *The Myths of Tattoo Removal*

1. *It's fast*
2. *It's painless*
3. *It leaves skin like new*
4. *It works for all colors*
5. *It works for all patients*

laser energy may make the inflammation that much worse. That is why there needs to be an option to excise the tattoo with minimal heroics such as skin grafts. On several occasions we have surgically excised residual areas of green or other colors once treatment of the rest of the tattoo has been optimized. A bigger tattoo may be removed with staged excisions, with or without tissue expanders, but these scars always look like someone has gone through a window.

Keep it in a place easily covered by normal clothing. By this I mean clothing normal for that gender and climate. People will make value judgments and sometimes these people may have greater influence over one's life than one's peers who have cheered you on. Men should not have tattoos below the upper deltoid; women should avoid the shoulders and arms completely. No one should get the face, neck, or hands tattooed and avoiding the legs between the bottoms of shorts and the tops of socks is wise as well. One may always choose clothing to display certain tattoos but many employers (including the US Army and US Air Force) prohibit art that cannot be easily covered with normal clothing or uniforms. For the potential tattoo wearer who fancies themselves exotic warriors, they should keep in mind that many law enforcement agencies and special operations groups prohibit unique, visible, and/or identifying tattoos. Even Sean Connery kept his "Scotland Forever" tattoo under heavy makeup or out of camera view until the much older James Bond of "Never Say Never Again". This rule also dovetails with the one above in that a tattoo which, for whatever reason, must be removed will leave a scar in a less obvious location.

No names. The reasons for this are obvious. Tastes, times, and

people change. If you must get a name other than your own or your mother's, get it small and in black so we may attack it more easily with our lasers later on.

No violent, racist, satanic, sexual, or obscene art. Not everyone wears or enjoys tattoos and one never knows for whom one may want to work for some day. New employment, or in some cases continued employment, may be dependant upon the successful removal of an offensive tattoo. What is "offensive" is not determined by the wearer of the design but by external and sometimes powerful forces. This sometimes forces the patient into having to make the uncomfortable choice of undergoing a rapid but disfiguring surgery or seeking a job elsewhere. This may keep you out of the armed services and may certainly get one into fights with equally violent or offensive people. If you would not want your mother, your boss, your co-workers or your children to see it, it shouldn't be put on in the first place.

Don't get it while intoxicated. This is obvious. People make bad choices while drunk or stoned and may end up with a large, multicolored swastika adorned with the name of the girlfriend who just dumped them on their neck. It seemed like the thing to do at the time.

The above are all reasonable things to bring up with the teen that is entering the tattoo vulnerable age. I've told my own teenage boys that I won't stand in their way if they simply MUST get a tattoo and that I will even pay for it. Part of the deal, however, is that I get to choose where it's performed and have veto authority over certain aspects of color, size, design, and body location. That took all the fun out of it and they haven't called my bluff (yet).

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## Community Acquired MRSA in Children: An Emerging Clinical Problem

Julia Lynch LTC MC  
Deena Sutter Capt MC  
Mike Rajnik Maj MC  
Pediatric Infectious Disease  
San Antonio  
Military Pediatric Center

### Introduction

**R**eductions in HIB and Pneumococcal associated disease has left *Staphylococcus aureus* (SA) an increasingly important cause of serious bacterial infections in children. It is responsible for >80% of bone and joint

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Table 1.  
Infections Associated with *Staph aureus*

- Superficial Infections
  - **Impetigo**
- Deep Skin Infections
  - **Furuncles, Carbuncles, Cellulitis, Wound Inf.**
- Respiratory Tract
  - **Sinusitis, Pneumonia**
- Musculoskeletal
  - **Osteomyelitis, Pyomyositis, Arthritis, Diskitis**
- Head & Neck
  - **Cervical Adenitis**
- Cardiovascular
  - **Endocarditis, Sepsis, Thrombophlebitis**
- Visceral Abscesses
  - **Liver, Renal**
- Foreign Bodies
  - **Vascular grafts, shunts, hardware**

infections, the majority of skin and wound infections, as well as infections in a wide range of other locations (table 1).

Although common in adult populations, methicillin and other drug resistance among SA has been rare in children without risk factors like exposure to an ICU or chronic health care facility. However, multiple recent reports suggest the emergence of a new variant of methicillin resistant *Staphylococcus aureus* (MRSA) among children without traditional risk factors. This community-acquired MRSA (CAMRSA) is distinct from nosocomial MRSA in a number of ways. The increasing prevalence rates of these organisms in our community should affect a clinician's empiric choice of antibiotics when a SA infection is suspected.

### Staph aureus and antibiotic resistance

Prior to the introduction of antibiotic therapy, mortality due to SA bacteremia was greater than 90%. When Penicillin G (PCN G) became available, the prognosis for a SA infection was greatly improved. However, by 1942 PCN G resistance att

makes the PCN inactive) was documented and spread rapidly from hospital to community such that currently less than 10% of SA isolates are susceptible to PCN G and this agent is not considered appropriate therapy for a SA infection.

Chemical modification of the

penicillin molecule led to steric protection from the b-lactamase and thus the success of the anti-staphylococcal penicillins commonly used in practice since the 1950's- methicillin, oxacillin, nafcillin, dicloxacillin. Addition of a b-lactamase inhibitor like clavulanic acid or sulbactam to amoxicillin or ampicillin has also been effective at getting around the penicillinase that most SA produce.

Methicillin resistance, however, occurs by a distinct mechanism not related to b-lactamase enzyme production. Methicillin resistance is conferred by the chromosomal mec A gene that encodes an altered penicillin binding protein (PBP-2A)- the target molecule for PCN in the bacterial cell wall. This altered protein prevents all b lactam antibiotics (PCN's and cephalosporins) from interfering with cell wall synthesis. Thus methicillin resistance equates with b-lactam resistance and there are no effective inhibitors of this process available for use in humans.

### Nosocomial MRSA

Infections due to MRSA first emerged in hospital settings but have become an increasingly large problem in the US. In 1974, 2% of the nosocomial isolates were methicillin

resistant. By 1997 the prevalence had increased to fifty percent. MRSA also "spread to the community" primarily among adults, but remained generally associated with specific risk factors that included recent or frequent hospitalization, residence in a long-term care facility (LTCF) or use of illicit intravenous drugs. Both of these latter risk groups generally also had extensive exposure to hospitals and the MRSA strains identified among these groups are similar in drug resistance patterns and genotypic pattern to the hospital acquired or nosocomial MRSA's.

In addition to methicillin resistance, greater than 50% of these nosocomial origin MRSA isolates also have resistance genes (linked on the piece of chromosome with the mec A gene) to numerous other antibiotics (erythromycin, clindamycin, TMP/SMX) and thus are often only reliably susceptible to vancomycin. Therefore, for suspected hospital acquired SA infections, vancomycin has been the empiric drug of choice until susceptibility testing can be accomplished. Vancomycin is an expensive drug with a significant toxicity profile. Excessive use of vancomycin is discouraged because of fear that it will drive development of resistance in other bacteria like *Enterococcus sp.*

### Emergence of CA-MRSA

In spite of the increasing prevalence of MRSA among adults in the US, the SA isolates among children have largely remained methicillin susceptible community acquired organisms and thus empiric antibiotic treatment for suspected SA infections has safely been a b-lactam antibiotic like methicillin for the last several decades. However, since the late 1990's community acquired MRSA strains have been increasingly identified among

children treated at hospitals in many cities in the US including cities in Texas. In fact, Driscoll Children's hospital in Corpus Christi reported nearly 50% of all SA isolates from children in 2001 as CAMRSA. Few of these children had any known risk factors for acquiring MRSA (no recent hospitalization, surgery, exposure to a resident or employee of a hospital or LTCF).

SA is commonly carried asymptomatically in the nares of about 30% of all children. Studies done in the last several years to assess the prevalence of CAMRSA carriage among children in the US have found from 1-10% nasal carriage rates.

**How are these MRSA different?**

Genetic analysis of these CAMRSA isolates indicates that these organisms are "highly clonal" or very closely related to each other and quite distinct from nosocomial MRSA isolates within the same community. The methicillin resistance is by the same mechanism- the mec A gene. However the genetic element or piece of chromosome containing the mec A gene found in the CAMRSA is distinct and less often contains other drug resistance genes. Therefore, it is clinically important that CAMRSA have a much lower association with resistance to other drugs, particularly clindamycin and TMP/SMX, than the traditional nosocomial MRSA.

A number of clinical reports on CAMRSA from children's hospitals also suggest a relatively higher association of these strains with skin and soft tissue infection rather than with other sites of infection. Reports from both Driscoll and Texas Children's Hospitals indicated that ~90% of their CAMRSA isolated from children were associated with skin and soft tissue infection- a significantly higher proportion than with other SA isolates. These clinical

observations are supported by the finding of the co-occurrence of a virulence factor for skin/soft tissue infections and pneumonia, known as P-V leukocidin determinant, with the mec A gene among patients infected with a CAMRSA in France.

**Are there risk factors for CAMRSA among children?**

Two studies have tried to assess for risk factors that could alert health care providers to be more concerned about CAMRSA in a child with an infection. All of the following potential risk factors were explored: age, race, socioeconomic status, recent antibiotic exposure, day care attendance, presence of underlying disease, previous hospitalization, number of health care visits, exposure to health care workers in the home. None of these have thus far been identified as reliable risk factors. Consequently clinicians are left with the reality that any child with a possible SA infection could have CAMRSA.

**How should this information change my practice?**

CAMRSA infection among children is a new reality that we must incorporate into our practice behaviors. An informal assessment of all SA isolates cultured from children in the WHMC microbiology lab over the last 2 years indicates that ~30% are now CAMRSA.

Several guiding principles

should now be applied whenever evaluating and treating a child in whom SA may be the infecting organism.

1) Culture all wound and relevant body sites at the time of presentation and before starting your first empiric therapy.

Obtaining cultures when faced with a serious infection has long been a practice standard. Now with CAMRSA, consider culturing even minor infections so that susceptibility testing can guide therapy. This will prove very useful if the infection is not adequately improving several days into your initial therapy.

There is an important twist to be aware of when using susceptibility results to guide therapy. Erythromycin and clindamycin are related antibiotics that share vulnerability to some but not all macrolide resistance mechanisms. SA isolates can be resistant to erythromycin alone or be resistant to both erythromycin and clindamycin. A particularly challenging occurrence is the phenomena of inducible clindamycin resistance seen among some of the erythromycin resistant isolates. In this circumstance the isolate will be reported as resistant to erythromycin and appear susceptible to clindamycin on initial lab testing, but will develop resistance to clindamycin on continued exposure (on therapy!). The inducible resistance trait can be detected when a microbiology lab performs a "D-zone test". Consequently, when a SA isolate is reported to be erythromycin resistant, the clinician should

*Table 2. Suggested Empiric Therapy for Staphylococcus aureus Infections*

<b>Vancomycin</b>	<i>Serious, life threatening and systemic infections ( sepsis, pneumonia, endocarditis, central lines, deep tissue infections)</i>
<b>Vancomycin</b>	
<i>Or plus Drainage</i>	
<b>Clindamycin</b>	<i>Moderately severe focal infections (osteomyelitis, septic arthritis, superficial abscess or cellulitis with systemic signs/symptoms like fever)</i>
<b>Clindamycin</b>	
<i>Or plus Drainage</i>	
<b>TMP/SMX</b>	<i>Mild infections (impetigo, superficial abscesses/furuncles without fever) Topical mupirocin- Impetigo only</i>

always request the lab check for inducible clindamycin resistance with a "D-zone test".

2) For serious life threatening Infections assume that CAMRSA may be involved and begin an empiric regimen that would provide adequate coverage.

Specific Recommendations for empiric therapy while susceptibility results are pending are seen in Table 2:

### **Is there no longer a role for b-lactam antibiotics in the treatment of SA?**

Many minor infections with CAMRSA in a normal host will get better on a b-lactam if adequate debridement/drainage occurs at the site of infection. We have many decades of experience to indicate that b-lactam drugs are safe and well tolerated in children. Therefore, they remain appropriate and even many times preferred drugs for SA once the isolate has been determined to be susceptible. Unfortunately the time has come when that can no longer be presumed, but must be proven.

### **Are there any new antibiotics that will give us additional choices in treating CAMRSA?**

Linezolid (Zyvox) is a new drug that has proven useful for treating antibiotic resistant Gram-positive bacteria like MRSA. It is an Oxazolidinone antibiotic that works at the 50 S ribosome in the bacteria to interfere with protein synthesis. There is still relatively little experience with this drug, particularly in children. One study in pediatrics noted an 80-85% cure rate in MRSA skin and soft tissue infections that had failed prior therapy. The major toxicity of the drug is bone marrow suppression resulting in thrombocytopenia. Among the drugs most

useful characteristics is excellent bioavailability when administered orally and availability in a suspension. The drug is very expensive and at present only available by consultation with an Infectious Disease provider.

### **Conclusion**

CAMRSA infections in children appear to be a problem unlikely to go away. Many researchers, including several residents and staff at SAMPC, are working on understanding the dynamics of this emerging clinical problem. In the meantime we must adapt our current practices to this new reality.

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secondly, am I doing OK as a parent? They need these reassurances from their provider and expect knowledgeable feedback.

Unfortunately, meeting these needs is not always the priority of the physician during a rushed well child check. Behavior, discipline, and anticipatory guidance take time to address effectively. The challenge becomes "How can I meet the needs of the parent within the time constraints of an already busy appointment?"

Using "Teachable Moments" is one strategy to provide effective education. It was developed by the Commonwealth Fund group in conjunction with Boston University School of Medicine and is the cornerstone of the Healthy Steps model for providing care to children and their families. It is based on the principle of using the basic components of the pediatric visit – the history, physical exam, and developmental assessment – as opportunities to provide education and insight into their child's development, temperament, and behavior.

This strategy can be easily incorporated into the visit in several ways: using the child's behavior and parental reaction in the office as a springboard for teaching, using

parent's questions, or actively creating teachable moments by eliciting comments about a child's behavior from the parent.

### **Observing and Interpreting Behaviors**

The basic premise is this: you observe a child's behavior in the office, comment on it in developmental terms, ask the parents how they feel about it, and reframe it in a positive light. Very often those things that a parent finds most frustrating and concerning (mouthing toys at 6mo, throwing blocks or food at 8mo, fierce independence as a toddler) are completely normal and developmentally expected. Using these witnessed shared events between the parent and provider provides a unique opportunity to further the parent's understanding about their child.

Let's take an example.

Mrs. Garza brings her 3yr old son for a well-child check. He is a very active child and climbs up on the examining table, stands up, jumps off and repeats. Again and again. You try to continue taking the history, glancing nervously over at Joey's gymnastic events, while Mrs. Garza remains seated. She frequently interrupts her conversation with you to yell, "Joey, get down. Stop jumping." "Joey, I said, NO jumping." "Joey, are you listening? I said, NO! Do you want a spanking?" Joey continues to climb and jump. Mrs. Garza's voice continues to get louder. This moment provides the perfect opportunity for you to discuss discipline with the mother. You've just shared her ineffectiveness at getting Joey to stop his undesired behavior. You comment how frustrating it must be to try to contain an energetic preschooler followed by asking the mother what types of discipline seem to work best with Joey. Do she and her husband

share the same philosophy on discipline? You note out loud that the mother's verbal command to stop the behavior without resultant consequences is unlikely to change Joey's behavior. He needs to learn that "no means no." You follow-up with a discussion about the effectiveness of time-out, "catch 'em being good", etc. You end the interaction with comments about how his energy and persistence will serve him well as he continues to master new skills. As you can imagine, discussing parenting strategies at this time in the visit is likely to be better received and implemented than as an afterthought at the end of the visit.

### **Creating Teachable Moments**

The physical exam also offers opportunities to incorporate teachable moments into the visit.

Let's take another example.

During the visit of a 6 month old, you are trying to listen to the baby's heart. She verbally protests and repeatedly tries to pull the stethoscope off her chest. You offer her a couple of tongue blades to hold in each hand and demonstrate how she can bang them together. She immediately tries to place one of them into her mouth. You are able to complete the chest exam without difficulty.

This brief exchange provides several opportunities for teaching. The parents have observed your ability to distract their child to allow you to continue with a job that the child was initially resistant toward. You have developmentally assessed the baby's ability to manipulate objects, and you can briefly comment both on her developing fine motor skills and babies natural propensity to explore their world by mouthing objects. Now that they have witnessed with you their baby becoming more accomplished in

reaching and grabbing objects, this interaction provides a nice transition to discussing the anticipatory guidance of needing to baby proof their house. Again, the guidance offered in this setting is more powerful and likely to be heeded than as one of many in a laundry list of requests of parents.

### **Using parental questions**

Your opening line of "so, how is everything going with Amy?" also provides opportunities for teachable moments. Amy's mother volunteers how 'clingy' her 9mo old child is, crying whenever she leaves the room. Consequently, she spends much of her day carrying the baby around. Her husband is concerned she is spoiling the baby. The mother's responses to your query offer the opportunity to discuss stranger anxiety in the developmental context of object permanence. In another visit, a father remarks that his sister's 22mo old daughter is potty-trained. He is concerned that his barely 2 year old daughter is not. This father needs to know it is OK that his daughter is not potty-trained – in fact, she is not expected to have achieved this milestone at her age. You review all the developmental milestones that his daughter has attained while reassuring him that his daughter is right on track with potty-training (her vocabulary is increasing, her gross motor skills are improving, she sits on the potty chair, etc). In those few moments, you have heard his fears and recognized his anxiety that his toddler may not be developing appropriately. Your reassurance may be the most important information you convey during that visit.

### **Eliciting Teachable Moments**

Often a single well child visit gives multiple opportunities for "teachable moments." However, should these opportunities not

present themselves during the visit, you can easily elicit them by asking pertinent, open-ended questions. “How is bedtime going?” “Any concerns about her eating?” Again, listening carefully to the parent’s answers can oftentimes reveal discrepancies between parental expectations of their child’s behavior or development and more realistic developmentally appropriate behavior (ie expecting a newborn to “sleep through the night”). It’s your chance to correct these misperceptions and provide parents with accurate knowledge or simply reinforce the positive things they are already doing.

### Modeling behavior

Finally, teaching by example can also be a powerful tool to enhance parental learning about their child’s behavior and how to handle difficult situations in a positive way. We model behavior every day as we interact with their children, and often it is this visual teaching that is remembered far more readily than our spoken words. Examples of this include limit setting in the office for a rambunctious toddler, helping to comfort a fussy 2wk old by demonstrating various rocking techniques, distracting toddlers away from our medical equipment toward more appropriate toys or offering a 6mo old a more appropriate toy to mouth (rather than the cord of our otoscope!), or engaging a 2yr old in reading a book – naming objects, asking colors, etc.

### Summary

Every parent has their own style of parenting just as every practitioner has their own style of “doctoring.” Incorporating “teachable moments” into your practice can be a powerful tool to enhance your effectiveness in providing education to your families in the reality of a busy office visit. I encourage you to

take the next step – spend a few moments to reflect back on your own last few appointments – think of “teachable moments” that perhaps slipped by and come up with ideas of how you can bring them into your future visits. While we will all use these teachable opportunities differently and to varying degrees of success, your parents will appreciate your continued commitment to them and their children.

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*eye on him and took him home when he cried.*

*We all grew up, moved on, and when I was home for Christmas from my freshman year at college, I found Stevie out on the playground and we talked. He was thrilled to see me. I think I was uncomfortable. For some reason I don't completely understand or recall, I asked him to come over to my house where we had some lunch and played pool for a while, a very odd experience for me. He was different than anyone I had ever met.*

*Stevie had Down syndrome.*

*A few years later, his mom told my mom how happy Stevie was that I had talked to him and spent some time with him. For my part, I was off-balance. What was going on with this person? I did not understand him. My uncertain grasp of human nature, of man's place in the universe, was wrapped up in literary criticism, William James, Noam Chomsky, linguistics, epistemology. Stevie seemed outside all of that. But why?*

Mental retardation is defined as significant impairment of cognitive and adaptive function, beginning before age 18, and often is first manifest as developmental delay. For some mentally retarded individuals, the underlying cause, as with trisomy 21, is straightforward, but for a large percentage of individuals with MR, ranging from 20 to 55%, we do not understand the cause. The Online Mendelian Inheritance in Man (OMIM) database, which excludes multifactorial, environmental, and chromosomal disorders, lists 1133 entities under MR. The London Neurogenetics Database, which focuses on syndromic conditions, has 1486 diagnoses that are associated with MR. A search of PubMed yields 60,463 “hits.”

For the primary care provider, a solution often does not seem easy. There is hope, though – diagnostic strategies, tools, and resources are accessible and have proven track records of success.

Launching into an elaborate diagnostic algorithm begs an important question: Why? Not “Why does this person have MR?” but “Why should I do this? What’s the point?” It is critically important to understand the answer to this question and to explicitly articulate to parents why you choose to seek an answer. This initial aspect of the diagnostic odyssey, though frequently overlooked, constitutes the

foundation for all subsequent investigation. It is, in fact, a contractual understanding between the care provider and the care recipient and should be worked out to mutual satisfaction. Here are some answers:

1) Establishing a precise diagnosis solves a mystery, often banishes the demons of guilt and blame, brings the life of a child out of the blackness and into the light of reason, and satisfies our human nature to understand causality. Parents usually want truth, even if we are powerless to change it.

2) With diagnosis comes prognosis. How will this child do in school, will she live independently, have children, grow old? Will we need to prepare financially for her long-term care? Will she have medical complications? What can we do to prevent them? For many diagnoses, knowledge of the natural history of the disorder has allowed us to formulate specific health care guidelines in which surveillance for hormonal deficiencies, cancer, behavioral issues, and organ dysfunction can improve quality of life and avoid premature mortality.

3) Diagnosis informs us about recurrence risk. Not only parents, but also siblings, aunts and uncles, and sometimes more remote relatives may be at increased risk for bearing a child with the same problem. If they are not at increased risk, this information can be of considerable

importance as well.

This aspect of medical care has some taint of eugenics, as if one of the aims of genetic diagnosis is to prevent further occurrences, to control reproduction on the basis of supposedly “scientific” values that serve a social agenda. For some practitioners and parents, the mere mention of the term “prenatal diagnosis” suggests we are potentially leading couples down the road to consider abortion. This is neither the intent nor the reality of recurrence risk counseling. The other side of the coin is embossed with the ethical principle of autonomy: If we come to an understanding of recurrence risk, who are we to withhold this information from the individuals to which it applies?

The incidence of mental retardation is estimated as 1-10 percent. Males outnumber females at a ratio of 1.5 to 1. Table 1 summarizes several recent studies and reviews that categorize various etiologies of MR. As you can see, chromosomal aberrations constitute the single largest category of MR in which the cause is known. Metabolic conditions, though important, are relatively infrequent. Single gene disorders and syndromic conditions are also important causes. In Table 2 are listed the top 10 single gene disorders ascertained in South Carolina over the past two decades.

In the past ten years we have discovered that a significant proportion of MR is caused by very small chromosomal deletions, duplications, or translocations in the subtelomeric regions.<sup>3</sup> These areas just beneath the tips of chromosomes are particularly rich in both genes and tandemly repeated segments of DNA, which predispose these areas to clinically significant rearrangements during meiosis. The good news is that these rearrangements are readily detectable using fluorescent in situ hybridization (FISH) and other molecular techniques and will disclose an abnormality in as many as 10 percent of individuals who have had exhaustive, but negative, traditional workups. The bad news, at least for now, is that these techniques are relatively expensive, on the order of about \$1000 per specimen. Consequently, several research groups have suggested that subtelomeric assays be utilized as a second-tier test. Retrospective analysis indicates that the following features will help select patients more likely to have a positive subtelomeric assay: a family history of MR, poor prenatal growth, decreased or increased postnatal growth, two or more dysmorphic facial features, and one or more non-facial dysmorphisms or congenital anomalies.

The primary care provider is in an ideal position to orchestrate the evaluation of a child with MR. Following screening and confirmation of MR, one does not have to automatically relegate all of the subsequent investigation to the developmental pediatrician, child neurologist, or geneticist. Though these consultants are important allies and should be part of the team effort, they are often neither immediately nor frequently accessible. The fundamental clinical tools and laboratory tests are well within the purview of the generalist. These include:

Table 1. Causes of MR

Cause	Stevenson et al <sup>1</sup>	Battaglia <sup>2</sup>
Unknown	56%	25-38%
Chromosomal	11	4-34
Subtelomeric rearrangements		7
Single Gene	8	
Fragile X	1.7	
Culturofamiliar	6	
Injury	5	
Infection	5	
Prematurity	5	
Metabolic		0-5
CNS Malformations		10-15
MCA Syndromes		4-5

Table 2. The ten most frequently diagnosed single gene disorders, in rank order (N=10,997)<sup>1</sup>

1. Fragile X syndrome
2. Other XLMR conditions
3. Tuberous Sclerosis
4. Angelman Syndrome
5. Williams Syndrome
6. Neurofibromatosis type 1
7. Prader-Willi Syndrome
8. Rett Syndrome
9. Myotonic Dystrophy
10. Phenylketonuria

1. A complete history, beginning with prenatal maternal health, medications, teratogenic exposures, and test/ultrasound results, thorough perinatal/birth history, and meticulous developmental history. A careful review of records is imperative.

2. A three-generation pedigree.

3. A thorough physical exam, with measurement of head circumference and comparison to norms, and careful attention to dysmorphic features. Examination of the parents is highly recommended.

4. Audiologic and ophthalmologic evaluations. Occult deafness and ocular pathology can serve as

excellent “handles” for pursuing a diagnosis.

5. CNS imaging, by MRI preferably, especially if the head circumference is greater than the 95<sup>th</sup> or less than the 5<sup>th</sup> percentile. In these cases, parental OFCs should also be measured.

6. Karyotype (chromosome) analysis and fragile X DNA analysis.

7. If the clinical scenario suggests an inborn error of metabolism, screening metabolic labs, to include electrolytes, glucose, lactic acid, ammonia, hepatic enzymes, urine organic acids, and plasma amino acids.

A number of algorithms for the evaluation of mental retardation are available, notably in Rudolph’s Pediatrics, the text we now use for resident textbook reviews here at BAMC. Many practitioners find these diagrammatic guides quite useful, yet one can do little wrong by beginning – and returning — to the basics: history and physical. The eminent dysmorphologist Bryan Hall, though, has articulated perhaps the most lustrous pearl of wisdom:

“Follow-up is the diagnostic ace in the hole.”<sup>4</sup> Pursuit of a diagnosis, like all good clinical care, is most routinely a matter of seeing the patient serially over time. No big job gets done all at once, and in the evaluation of MR, often a critical clue or a critical insight takes time to develop.

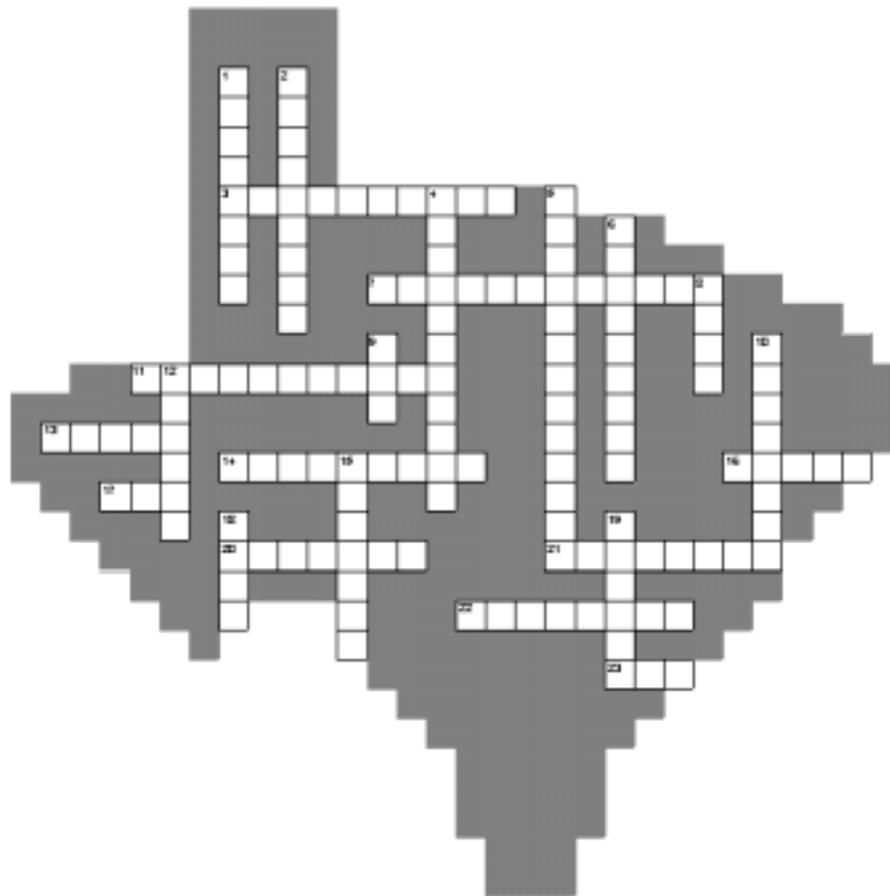
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The information and opinions stated in the *Pediatric News* are the opinions of the authors and in no way reflect official policy or medical opinion of the United States Army or any other government agency.

John Baker, M.D.  
 Editor, *Pediatric News*  
 Department of Pediatrics  
 San Antonio Uniformed Services Pediatrics  
 jabaker@texas.net

## February 2004 Crossword Puzzle



Word List

behavior  
black  
chromosomal  
connery  
creating  
fragilex  
hiv  
isotretinoin  
linezolid  
modeling  
none  
observe  
prognosis  
sarcoidal  
scar  
staph aureus  
subtelometric  
ten  
twelve  
vancomycin  
white  
why  
yellow

Across

3. this antibiotic has been the empiric drug of choice until susceptibility testing can be accomplished for suspected hospital acquired staph aureus infe
7. tattoos and piercings should not be obtained while patient is on or has recently completed a course of this drug
11. the organism responsible for greater than 80% of bone and joint infections in children
13. tattoo color that turns an ugly gray with laser treatment
14. granulomatous inflammation associated with tattoos
16. current laser technology for tattoo removal works best on these pigments
17. there has never been a documented case of this transmissible disease through a professionally placed tattoo
20. actor with "Scotland Forever" tattoo on right forearm
21. the process of \_\_\_\_\_ 'teachable moments' during the well child exam may be more powerful than a laundry list of anticipatory health requests for
22. the top single gene disorder identified as a cause for mental retardation
23. prior to launch into an elaborate diagnostic algorithm to determine the etiology of an individual's mental retardation, this wuestion must be asked

Down

1. besides development, often one of the key issues for most parents during a well child visit
2. one answer, which might provide valuable information relating to his future care, that is sought in attempting to obtain the reason for a child's men
4. these alterations constitute the single largest category of mental retardation for which the cause is known
5. because of the expense of testing with fluorescent in situ hybridization (FISH) this type of testing should be a second-tier test
6. a new drug that has proven useful for treating antibiotic resistant gram-positive bacteria like MRSA
8. of the potential risk factors for a child to have CAMRSA, the one found to be most reliable is \_\_\_\_\_
9. studies done in the last several years have identified up to \_\_\_\_\_ percent nasal carriage rate of CAMRSA in children in the U.S.
10. your interaction and appropriate limit setting with a child in your office is an example of \_\_\_\_\_ behavior
12. optimum laser treatment of a professionally applied black tattoo may take \_\_\_\_\_ monthly treatments
15. the basic premise of the "teachable moments" model is to \_\_\_\_\_ the child's behavior in the office
18. any quick method to remove a tattoo is certain to leave this in its place
19. another tattoo color that turns an ugly gray with laser treatment