Newborn Screening Changes: What Pediatricians Need to Know
Friday, February 3 Conference Call – Summary

Hosts: Marilyn Peitso, MD, MN-AAP president; Anne Edwards, MD, MN-AAP policy chair; Amy Gaviglio, genetic counselor at MDH; Joanne Bartkus, director of public health laboratories division at MDH.

Drs. Marilyn Peitso and Anne Edwards: Thanks to all who are participating in this call. As an organization advocating on behalf of children, we are concerned about the changes to the newborn screening program and want to make sure providers and families have the most accurate information available.

Joann Bartkus: I’m going to start with some background information to let you know how we arrived at these changes. Up until November 16, 2011, our policy had been to store residual blood spots and the data associated with those blood spots indefinitely unless a parent specifically requested that the information be destroyed. Parents have always had the option to opt-out of the screening itself and/or request that the child’s blood spot be destroyed. The blood spots that we saved were used for program operations, including quality assurance and improvement activities, and we made those spots available to researchers for selected studies.

In 2006 Minnesota State Statute 13.386 was passed, commonly referred to as the genetic privacy act, stating that collection, use and dissemination of genetic information was prohibited unless expressly authorized in statute. Minnesota statutes 144.125 - 144.128 authorize newborn screening, so we believed we had express authority. In 2007 we went in front of an administrative law judge to change our rules surrounding newborn screening and the ALJ at that point mentioned the conflict with 13.386. In 2008 we went to the Minnesota legislature and tried to resolve the apparent conflict between the two statutes. The legislation was passed by both bodies, but was vetoed by Governor Pawlenty. We have tried every year subsequently to get something resolved in statute, but that has not happened.

In 2009 there was a lawsuit filed (Bearder et all v. State of Minnesota) charging that the Minnesota Department of Health was violating 13.386 by storing residual dried blood spots and using them for purposes other than the actual screening. We had moved to dismiss that lawsuit and that dismissal was granted in district court in Hennepin County. The plaintiffs appealed that decision and the appeal was upheld by the Court of Appeals. However, on November 16, 2011, the Minnesota Supreme Court reversed that decision, ruling that the statutes for newborn screening did not give express permission to store the samples beyond the point of testing, to use those samples for anything other than the purpose of screening the baby, or to disseminate the results of the testing. The court interpreted “biological information” as meaning DNA. So they basically stated that 13.386 not only covered genetic data
derived from analysis of blood spots but also the samples themselves because they contained DNA. Since the ruling on November 16, we’ve had a number of discussions about what compliance means. We’re still in the process of trying to get legislation in place that will further clarify things. We proposed that we start destroying the blood spots after 71 days based on looking at data from a presumptive positive test result and confirmation. This is a temporary ruling. And data will be kept for two years as required by CLIA.

Amy Gaviglio: We recognize that 71 days has the potential to miss some cases and we know there are some disorders that take longer than 71 days to diagnose. What we’re hoping to do in order to retain the specimens of those cases is ask for consent to retain the specimens of any child with an initial positive screening result. What that means for pediatricians is that you’ll receive a consent form with the usual packet of information that goes along with a positive result that parents will need to sign. It’s also likely that you will have parents contact you and say they don’t want their blood spots destroyed. At present, we are not able to retain negative blood spots. However, they can request that the blood spots be returned to them through the physician. We recognize that this is a burden on you, but we don’t feel comfortable sending this back to parents directly. The process would be for the physician to fax us a request on behalf of the parents. We will then pull the specimen and send it back to the clinic so you can give it back to the parents when they come in.

Another question that seems to be prevalent is there seems to be a discrepancy between the new newborn screening brochure and the 71 days. The latest newborn screening brochure dated November of 2011 does not expressly state the 71-day retention period. It does discuss that parents have the option to ask for destruction. This is an option that parents still have even despite the 71 days. We encourage parents not to ask for this. We do ask that you continue to hand out that brochure as it covers a lot of important information. We also encourage you to direct parents to our website, which will have more up-to-date information on what their options are and the retention period. They can also call the MDH genetic counseling line at 651-201-3548 and speak with a genetic counselor.

The final question I will address is what impact this has on hearing screening as we recognize that this can take quite a long time to diagnose. The bloodspots are not used for hearing screening follow up. It’s only the data. Per CLIA regulations, the data will be stored for two years, which should cover the majority of hearing loss follow ups.

Q&A

Dr. Anne Edwards: As you talked, you said parents would be able to retain negative screens; however, my understanding is that the spots would not be of any future utility. Secondly, how will they be returned to the provider?

Amy Gaviglio: Parents can retain the bloodspots however they want at home. It still has potential utility, not for quality control or quality assurance purposes at MDH. As far as how the blood spots will be returned, it will be in a plastic bag inside an MDH envelope with a low biohazard risk sticker on it to your office. At that point you can work with parents on the best way to get it back to them, whether they want to wait for a specific clinic visit or come in and pick it up.
Dr. Marilyn Peitso: Are there environmental requirements for storing these blood spots? Are there situations where the quality would deteriorate and its utility would suffer?

Amy Gaviglio: Yes, environmental changes can have an effect. Generally speaking, we would say keep it dry and in a freezer.

Joann Bartkus: We are working on legislation to try to fix some of this and make it easier on providers and us. We are working on a process where the consent will likely go on the blood spot card itself and get physicians out of the middle. This really is a temporary fix until we come up with something that’s a little more palatable. We don’t anticipate that there will be a lot of requests, but we want to give parents at least some options for getting their child’s blood spot back. This is the best way we could think of to do that.

Dr. Marilyn Peitso: I have some concerns about the information given to parents about this process. There could be a lot of confusion and liability for pediatricians, family practice providers and hospitals having what’s supposed to be official information that is incorrect. Perhaps it would be helpful to have supplementary information to go along with the brochure that more explicitly outlines the process.

Joann Bartkus: I think that’s an important concern for us to take to our leadership and consider how to address that.

Dr. Jason Rausch: I’m a pediatric immunologist. One of the things that was tentatively supposed to begin this year was adding Severe Combined Immune Deficiency to the screening. Would this ruling delay that?

Amy Gaviglio: Part of the reason why it has the potential for being delayed is that our testing method for SCID is a completely new test. As part of being a CLIA lab, we need to validate our testing methods before we can add a test and report clinical results. When the ruling happened, we were not done with the validation of the test and we can no longer use blood spots for anything other than testing the newborn, which means we cannot use them for validation. Our solution for this is to look at obtaining specimens from another state. Michigan has a bio-bank of blood spots where parents have given consent for them to be used for purposes like this. So we have a way to get the specimens, but it will cost more money and take more time.

Dr. Jason Rausch: I can imagine that the validation process is ongoing. How does this impact future tests?

Amy Gaviglio: That’s a great question. CLIA requires ongoing validations, quality assurance and quality control and that’s one of our biggest concerns with this ruling. I believe our lab has done some estimates on how often they have to do validations and it was around three to four times per year. Those are validations, not counting standard quality assurance, so it’s a big concern.

Dr. Jason Rausch: So is this a done deal or is there anything we can do to change this?
Joanne Bartkus: We’re working on legislation but this is not a good political time. There is not a lot of movement that we see. We understand that this is very much a hot-button issue for the majority party. In the meantime, a worse-case scenario is that we’re going to have to get consent to store and use the blood spots. We’ll try to make that as easy as possible, perhaps by putting it on the back of the blood spot card, but there are a couple of problems with that. One is that it’s expensive and time consuming. Second, it’s not population-based. We will lose and possibly introduce bias into our validation in any work that we do with those spots. If there are studies that require populations that are perhaps underrepresented in those stored samples, we’re going to have to find a way to recruit from those populations. For those who have any interest in contacting their legislators, you’re certainly welcome to do that.

Dr. Anne Edwards: This is something that’s very important to families and we need to continue to look at all avenues to support this program and ensure that it remains as strong as it was in the past. We would like to hear from members and will be reaching out for individuals to provide feedback about concerns, so stay tuned.

Dr. Marilyn Peitso: Earlier the 71-day limit was discussed as being somewhat problematic. I wanted to raise the concern that on the clinic end, once we get notified, there may be problems getting the family in or contacting the family. I think the 71-day limit can be a real barrier from our perspective.

Amy Gaviglio: We recognize that and we’re re-examining what the maximum time has been over the last three years to confirm diagnosis and include everything you mentioned. We’re not happy about the 71-days either, but it ended up being a compromise. There are some people who feel the 71 days is far too long.

Joanne Bartkus: The plaintiffs did not object to 71 days for the positive samples. They objected to the 71 days for all of the samples. There’s a good rationale for keeping all of the spots for a certain length of time for administrative purposes. Certainly it was brought up that other states keep their blood spots for only 30 days, such as South Dakota, Oklahoma and Kansas. South Dakota doesn’t do their own screening; they send their samples to Iowa. So we were able to defend the 71 days, but there was definitely pushback. But I think we do have opportunity to lengthen that, especially for positive. We might carve out that exception. I think there’s a feeling that the Supreme Court ruling left us some wiggle room regarding presumptive positive samples.

Dr. Jason Rausch: Is there a contact person in our legislature who may be able to address why this is such a hot-button issue in the majority party?


Dr. Yeng Yang: When and if the blood spots are returned to the parents, are you going to be providing storage information or some kind of education along with that?
Amy Gaviglio: We certainly can include something about appropriate or best-case scenario for storage conditions. It’s good to have that suggestion before we start getting requests.