Emergency preparedness for newborn screening and genetic services

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Disclaimer: This guideline is designed primarily as an educational resource for medical geneticists and other health care providers to help them provide quality medical genetics services. Adherence to this guideline does not necessarily assure a successful medical outcome. This guideline should not be considered inclusive of all proper procedures and tests or exclusive of other procedures and tests that are reasonably directed to obtaining the same results. In determining the propriety of any specific procedure or test, the geneticist should apply his or her own professional judgment to the specific clinical circumstances presented by the individual patient or specimen.

Abstract. Patients identified in newborn screening programs can be among the most vulnerable during a disaster due to their need to have prompt diagnosis and medical management. Recent disasters have challenged the ability of newborn screening programs to maintain the needed continuity during emergency situations. This has significant implications for the newborn screening laboratories, the diagnostic confirmation providers, and the patients who either require diagnosis or maintenance of their therapeutic interventions. In 2007, the National Coordinating Center (NCC) for the Regional Genetics and Newborn Screening Collaboratives (RCs) sponsored a meeting involving representatives of the Regional Genetics and Newborn Screening Collaborative Groups, state newborn screening programs, providers of diagnosis and confirmation services, manufacturers of equipment, medical foods, and other treatments used in patients identified in newborn screening programs, and individuals from agencies involved in disaster response including the National Disaster Medical Service, the Centers for Disease Control and Prevention, the Emergency Management Assistance Compact, the Federal Emergency Management Agency, and others. In addition to developing contingency plans for newborn screening, we have considered other uses of genetics as it is used in DNA-based kinship identification of mass casualties. The meeting resulted in the description of a wide range of issues facing newborn screening programs, provider groups, and patients for which emergency preparedness development is needed in order that appropriate response is enabled. ***Genet Med* 2009:11(6):455–464.**

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During the past 10 years there have been numerous disasters and emergencies that have demonstrated the need to develop highly focused programs to ensure the continuity of health care and related services for patient populations and the public. Some emergency situations offer a narrow window of opportunity during which those likely to be impacted can prepare (e.g., Hurricanes Katrina and Rita)[1] whereas others occur with no warning (e.g., earthquakes and terrorism-related events). To ensure continuity of critical programs, emergency preparedness planning and ongoing assessment of the plans are critical. The public health and medical genetics communities recognize that some of their programs (e.g., newborn screening) and patients (e.g., those on critical therapies) are particularly vulnerable during these situations and that the technologies of medical genetics can be of importance in mass casualty situations.

Newborn screening detects a number (>29) of conditions including 22 inborn errors of metabolism, two endocrinologic, three hematologic, and two genetic conditions. These are single gene disorders, usually autosomal recessive that inactivates the function of a specific enzyme critical in intermediary metabolism. The results are often catastrophic: life-threatening coma due to hyperammonemia, severe acidosis, seizures, vomiting, and variety of other symptoms. They have in common the need for special diets that limit the intake of the offending food element while providing enough to permit growth and development. Many of the conditions are manifested in the first weeks of life, hence the need for rapid diagnostic confirmation of the putative positive newborn screen, and institution of appropriate diet therapy.

The development of newborn screening programs and the power of genetic technologies raise important considerations in the development of emergency preparedness plans.

Patients with genetic diseases who are identified through newborn screening programs are among the most vulnerable populations in that they are placed on therapies at birth to avoid the often high mortality and morbidity associated with their disease, if untreated, and this treatment must be maintained throughout life, without interruption, to avoid potentially catastrophic consequences of some of these metabolic diseases. In addition, many genetic technologies are fundamental to establishing the identity of individuals who may be involved in mass casualty events.

In August 2005, Hurricane Katrina brought home the need for organized disaster preparedness plans specifically directed at newborn screening. In the immediate aftermath of the storm, with the Louisiana Public Health Laboratory unusable, its chief had established the newborn screening program as one of the

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top three priorities due to the fact that every newborn throughout the state participates, births would continue to occur and irreparable harm could occur to infants if metabolic diseases were not detected in a timely way. Through a combination of fortuitous circumstances and activity, the newborn screening laboratory of the Iowa Public Health Laboratory was able to rapidly assume responsibility for the screening of Louisiana's newborns. Subsequently, the general lack of preparedness of most newborn screening programs led the Association of Public Health Laboratory's (APHL) Newborn Screening and Genetics in Public Health Committee (NSGPH) to initiate a major effort to define the critical elements of an emergency newborn screening contingency plan. Much of what was developed focused on the state program and its immediate responsibilities to perform timely screening and to ensure that infants with positive screens are referred into the private sector for diagnosis and management. More work remains to be done to develop contingency plans for this group of providers and patients.

In February 2007, representatives of state public health programs, federal agencies, disease support groups, hospitals and medical centers, device and therapeutics manufacturers, and providers of genetic services were convened by the NCC for the RCs to determine how best to meet the needs of patients with genetic diseases during disasters. The primary goals of the meeting were to identify many steps in the process of newborn screening, diagnostic confirmation, treatment, and management of the 4.1 million newborns each year in the United States and to determine how best to ensure the continuity of care for those newly identified, and for the existing patient populations in these categories during emergency situations. The full range of emergency management including preparedness, response, and recovery were covered with a focus on how best to mitigate problems that arise at each level. Most of these diseases are very rare and care for them is often complex. Trained and experienced providers are few. Care for conditions, such as the metabolic diseases, that are a large proportion of those to which newborn screening is targeted are often complex. Hence, successful logistic efforts to coordinate the newborn screening patient populations would be replicable to any patient with rare genetic disease requiring continuity of care during disasters.

Newborn screening is a program that includes more than just the screening event itself. Components commonly considered are (1) education of professionals and the parents of newborns who are to be screened; (2) screening, including specimen collection, submission, and testing; (3) follow-up of abnormal or unsatisfactory newborn screening results; (4) diagnostic confirmation of those identified in the screening program; (5) treatment and long-term management of the individuals diagnosed with one of the various diseases; and (6) program quality assurance and evaluation. All of these processes must be maintained for particularly time-critical diseases.

There are many participants in newborn screening programs including the public, state public health departments, primary care and specialty care professionals, and the institutions within which many of them operate. Hence, the steps in the process were considered from the perspectives of (1) state public health departments that are charged with the actual screening and notification of families and providers of the identification of a screen positive newborn; (2) medical institutions and providers that are charged with diagnosis and management; and (3) health interests of the families and patients who can participate to some degree at many stages of the program.

DISASTER RESPONSE ORGANIZATIONS AND AUTHORITIES

Many state and federal organizations assume specific responsibilities during a disaster situation. Local authorities tend to be in charge of managing the response and identifying outside resources to support the response. The authority under which they operate is commonly through entities that have the legal authority to allow exemptions to established practices and the power to mobilize necessary processes. First response to a disaster is a task for local emergency services providers, assisted by neighboring communities, the state, and volunteer agencies. In catastrophic disasters, after the request of the state's governor, federal resources can be mobilized through the Department of Homeland Security's Federal Emergency Management Agency that addresses many basic human needs such as food, water, shelter, and search and rescue. Because it typically takes 24–48 hours to bring nonlocal resources to bear, the Federal programs are least able to address local issues in the earliest stages of the disaster. Hence, local communities and individuals must ensure that they have adequately prepared for the emergency situation. A general axiom of emergency preparedness is that those who are least able to cope due to limited resources are the most affected.

Federal

National Disaster Medical Service

The National Disaster Medical Service (NDMS) is charged with "developing systems and plans to ensure that sufficient medical personnel, supplies, equipment, and facilities will be available and deployed to meet essential civilian and military health care needs during (national security or domestic) emergencies."[2] It operates out of the US Department of Health and Human Services (HHS). The NDMS is supplemented by state and local medical resources during disasters or major medical emergencies and provides back up medical support to the military and the Department of Veterans Affairs care systems during overseas conflicts. Working with HHS, the Department of Homeland Security, the Veterans Affairs, and the Department of Defense, NDMS provides medical response, definitive care, and patient evacuation. Medical response is led by HHS that coordinates the Disaster Medical Assistance Teams (groups of intermittent federal employees who volunteer to be on a designated team for NDMS). Teams of 35 persons with a range of health and medical skills are typically deployed for 2 weeks at a time before being replaced. The teams originate from community-based health and medical organizations. There are a number of specialty teams (anesthesiology, critical care, emergency medicine, family practice, general surgery, infectious disease, obstetrics and gynecology, orthopedics, pediatrics, and pulmonary care). Presently, there are no medical geneticists and/or metabolic disease physicians associated with NDMS and efforts are being made to find appropriate ways to involve them with the teams. NDMS is activated by the secretary of HHS or a designee. Federalization of the program resolves important issues such as licensure, certification, liability, compensation, and coverage under the Uniformed Services Employment and Reemployment Rights Act that also addresses issues of leave from employment and reemployment.

As relates to newborn screening and genetics patients, there are two pediatric teams and additional generalist teams in NDMS. Pediatric teams mostly include generalist pediatricians with limited experience in the management of patients with genetic diseases. NDMS is open to identifying appropriate ways

to involve clinical geneticists in their work and to develop systems through which point-of-care specialist support can be provided. There are also 10 pharmacist teams that can be involved in ensuring that patients are able to access critical therapeutics during emergencies. However, these would require considerable system support related to the organized stockpiling of the therapeutics, access to medical records, or treatment history and specialist support that are discussed later.

Disaster Mortuary Operational Response Team

The predecessors to Disaster Mortuary Operational Response Team (DMORT) arose in the 1980s in the private sector as a means of standardizing the approach to managing mass casualty events. After a number of airline incidents in the 1990s, DMORT became available as a federal response team. It has not had significant involvement in the area of newborn screening and genetics aside from having worked with mortuaries in the collection of specimens from a small subset of casualties that could be used in DNA identification.

PREVIOUS DEPLOYMENTS OF GENETIC PROFESSIONALS

The medical genetics community has been involved after several mass casualty events, including the World Trade Center bombing and hurricanes, though typically on an ad hoc basis. Prominent members of the genetics community served on the World Trade Center Kinship and Data Analysis Panel.[3] After Hurricane Katrina, genetics professionals served not only on the Hurricane Victim Identification Group that advised the Louisiana State Police and the Incident Medical Commander but were also recruited through personal connections and requests via listservs to volunteer in the Family Assistance Center supporting the DNA identification efforts. These volunteers greatly facilitated the DNA identification process as they were well prepared to work with distraught families to construct pedigrees for use in kinship analysis.[4] Genetics professionals have the unique training and expertise needed to elicit accurate family relationships when individuals are emotionally volatile and to identify family members who are informative for accurate kinship analysis. During the 3 months after Katrina, nearly 10,000 missing persons' cases were filed and over 200 bodies needed to be identified. Seven months after funding for DNA analysis was made available, 153 DNA reports had been issued and fewer than 40 bodies remained unidentified. Efforts toward formal integration of the genetics community and the standardization of the activities are discussed in the following article.[5] The volunteer geneticists played a significant role in the DNA identifications by locating missing people and reuniting families, identifying which family members to collect samples from[6] and gaining the trust of family members by listening to concerns, providing information on the identification process in a compassionate manner and managing expectations about the identification process.[5]

Centers for Disease Control and Prevention

The Centers for Disease Control and Prevention (CDC) is involved in emergency preparedness through their Coordinating Office for Terrorism Preparedness and Emergency Response (which provides a platform for public health emergency response). CDC also supports a Clinician Outreach and Communication Activity that establishes partnerships with national clinician organizations to communicate information about emergency and disaster events.

CDC takes an active role in quality assurance programs for state newborn screening programs and works with the APHL in that regard. Specific planning for a CDC role in emergency planning for newborn screening and genetics patients is in development in response to mandates in the recently signed federal Newborn Screening Saves Lives Act of 2007 (Public Law 110-204). Potential roles that have been discussed with the CDC include:

- Providing a backup for state newborn screening computer records,
- Developing a backup laboratory that can assume screening responsibilities for states during an emergency,
- Housing a backup pharmacy for the critical therapeutics required by patients identified by newborn screening as a part of the Strategic National Stockpile, a national repository of life-saving pharmaceuticals and medical supplies, and
- Incorporating clinical geneticists and metabolic disease physicians into the Public Health Information Network as genetics and newborn screening patient subject matter experts.

States

Emergency Management Assistance Compact

Emergency Management Assistance Compact (EMAC) is a Nation-wide Governors' interstate mutual aid compact that facilitates the sharing of resources, personnel, and equipment across state lines during times of disaster and emergency. EMAC is formalized into law by the member parties. It is governed by the National Emergency Management Association. EMAC maximizes the use of all available resources, coordinates deployment of EMAC resources with National Response Plan resources, expedites, and streamlines delivery of assistance between member states, protects state sovereignty, and provides management and oversight.

The EMAC process is engaged when a governor issues a state of emergency notification. Once activated, assessments of resource needs are made, cost issues are negotiated, formal requests are made for assistance from other states, and resources are deployed. Much of the success of EMAC is based on: (1) capacity to address issues of reimbursement, licensing, and liability at the time of activation that protects individuals who are involved in another state's activities; (2) prior development of Continuity of Operations Plans (COOP); and (3) continual improvement based on training and mock-disaster exercises. Depending on the magnitude of the disaster, regional or national coordinating teams may be engaged.

Individual states

The states are variable in the organization of their emergency response programs and the degree to which they address issues related to newborn screening and genetics patients. Much of the variability arises from differences in the types of program services that are contracted out or retained in state agencies and laboratories, the extent to which programs actively include short-term follow-up and long-term follow-up of patients, and the most likely types of emergencies that a state might face. In general, the deeper a state's history of having to deal with emergency situations, the more developed is its contingency planning. For instance, the state of California Department of Health Services' Genetic Disease Branch has developed detailed emergency planning and backup systems and has organized its system to provide redundancy through regionalization.

Unfortunately, most states are more like Louisiana in having only a single newborn screening laboratory available to them. Further, few states have the capacity to absorb the volume of another, particularly when the state in need screens for conditions that may not be screened in the contingency laboratory.

Regional genetics and newborn screening collaboratives

The Health Resources and Services Administration's Maternal and Child Health Bureau, through its Heritable Disorders Program, has funded a national system of RCs with an associated NCC to improve access to specialty health care services within local communities and to improve the integration of public health, primary care, and specialty care. Many of the RCs have developed specific programs that address various aspects of emergency preparedness. The New York - Mid-Atlantic RC gained first-hand experience in the needs of their region after the terrorism events at the World Trade Towers of September 11, 2001. Specimen delivery was compromised by the loss of some mail/package delivery services. The irradiation of packages led to specimen problems. Preceding this disaster was a 1993 electrical fire at the laboratory that shut down operations for 3 days. In response, extensive plans to backup the newborn screening laboratory were developed and issues related to program authorities (e.g., memos of understanding with backup facilities, funding, and ownership of specimens) were addressed. Operational roles and responsibilities and modes of communication were clarified.

As previously discussed, the Region 3 Genetics Collaborative in the Southeast was significantly impacted by Hurricanes Katrina and Rita. Communication between patients and providers and among providers was impaired such that access to the expertise of the highly specialized and limited number of metabolic disease specialists was a major problem. Again, first-hand experience led to the development of emergency management protocols with attention to the needs of local providers and institutions.[1]

The Region 4 Genetics Collaborative, which represents states bordering the Great Lakes and surrounding area, has developed a novel approach to ensuring that their newborn screening patients can access appropriate services during emergency situations. Although most metabolic disease physicians provide their patients with written information about what to do during a medical emergency, this RC has expanded the University of Minnesota Department of Pediatrics and the Midwest Emergency Medical Services for Children Information System Resource Center's web-based database that manages patient-specific Emergency Information Forms online. This program is expected to expand throughout the region next year. Further, they are working to link this system to their ongoing inborn errors of metabolism information system database through which they collect information about their patients' encounters with the health care system.

Professional organizations

Association for Public Health Laboratories

Although they may not do the actual testing, all state public health laboratories are involved with their state newborn screening program and hence are affiliated with APHL. After a series of natural disasters (floods) in 1992, the APHL addressed the issue of backup capabilities for state public health laboratories and their newborn screening programs through sessions at its annual meeting and its NSGPH. A subcommittee of NSGPH was formed to assess the status of contingency plans for US newborn screening programs and to provide guidance for establishing such plans. Recognizing that there are countless details involved in a workable plan, the subcommittee addressed the broad areas that must be a part of every contingency plan such as communication with hospitals, physicians, and parents; maintenance of testing, whether onsite or through contractual arrangements with another laboratory; provision of follow-up services; and provision for annual practice exercises of the system. APHL, in association with the National Laboratory Training Network, organized a webcast outlining steps necessary to establish a backup system for newborn screening programs. NSGPH has continued its interest in contingency planning, and is currently drafting a position paper for APHL on the subject. The major areas of concern for the testing laboratories are:

- Educating the family about newborn screening
- Collecting and transporting specimens
  - Monitoring to identify specimens that have not been received
- Shipping specimens to state screening laboratories
- Processing the specimens
  - Ensuring the availability of backup laboratories where needed
  - Ensuring the availability of reagents and equipment
- Reporting results to physicians and families, as appropriate
- Confirming diagnosis
  - Linking patients to appropriate medical providers
    - Accessing services and providers in other states, if necessary
    - Ensuring that medical emergency cases are able to access emergency services in appropriate facilities
  - Accessing the laboratories that provide diagnostic confirmation
  - Reporting those results to patients and providers
- Ensuring the availability of treatment and management resources either locally or at distant locations including:
  - Medical foods
  - Drugs
  - Emergency management

American College of Genetics

American College of Genetics (ACMG) operates the NCC for the RCs through a cooperative agreement with Health Resources and Services Administration/Maternal and Child Health Bureau. As the professional medical association that represents the board-certified clinical geneticists in the United States, ACMG is involved in the development of genetics-focused practice guidelines and clinical decision support tools for both specialists and primary care providers. ACMG is actively involved with national programs such as the American Health Information Community that has made newborn screening a use case in the development of an electronic health system capacity in the United States. This will allow newborn screening patients to be among the earliest of populations for whom clinical and laboratory electronic medical record languages are standardized and for which clinical decision support tools such as the newborn screening ACT(ion) Sheets have been developed. During time, many of the issues of newborn screening patients during emergency situations may be minimized by the availability of medical records in an electronic health care system.

EMERGENCY PREPAREDNESS PLANNING FOR NEWBORN SCREENING AND GENETICS

Emergency preparedness for newborn screening programs is predicated on the possibility of a disaster occurring that impacts individuals at any phase of the program, from obtaining and transporting their specimens to establishing of the diagnosis of those who screen positively to the treatment and management of those who are diagnosed. The state programs are in the best position to ensure that screening has occurred through their ability to match specimens to birth records and other vital records. However, once the patients move into the diagnostic and confirmation setting, local and institutional factors come into play with the state continuing to provide information to support the patients and the providers. At each step of the program, there are things that can be done by patients/consumers, institutions, and by the newborn screening laboratories and programs themselves to ensure continuity of programs and care. Although discussed in the context of newborn screening, patient with any genetic disease could be integrated into the planning once a diagnosis has been made. The three main functions include: (1) locating, developing, and deploying the resources to meet the various needs of the patients and programs such as medical care providers, functioning laboratories, and treatments; (2) empowering patients, providers, emergency preparedness organizations, and businesses to respond appropriately by having organized resources and plans that allow them to address the needs of this population; and (3) assisting patients, providers, emergency preparedness organizations, and businesses in accomplishing their tasks.

Newborn screening phase

In the first phase of newborn screening: (1) families/mothers are educated about the newborn screening programs before a blood specimen is obtained from their newborn; (2) the newborn's blood is placed onto a filter card, usually in the birthing facility; and (3) the filter card is forwarded to the screening laboratory for analysis. After analysis, the screening laboratory then (4) notifies providers and/or parents to initiate diagnostic confirmation of infants who screen positively.

There is wide variability among the states in the timing, content, and provision of general and more detailed education about newborn screening. Initiating education about the importance of these programs during the prenatal period may enhance a mother's and/or couple's knowledge about their involvement in the program in comparison with the education that commonly takes place in the hospital after delivery.

State programs should ensure the availability of filter cards for the collection and submission of specimens at all possible collection locations. The blood that is applied to the dried blood spot filter card, which also contains critical identification and contact information is typically obtained in the delivery hospital or birthing facility. However, this can also occur through midwives or in the office of the health care professional when deliveries take place in less organized environments. Once obtained, the dried blood spot filter card is transported to the laboratory responsible for screening. At the time of a disaster and specific to the screening phase, a patient sample may be vulnerable at any point up to receipt of the specimen by the screening laboratory.

If the newborn screening laboratory itself is not directly affected by the emergency, it should be able to process specimens that are or have been received and to initiate its own emergency COOP plan. If involved, the laboratory backup system is engaged. In either case, programs need to have mechanisms for tracking births and specimens that can be compared to ensure that all infants are screened. Tracking of missed infants or lost specimens may be required. Providers can also contribute by ensuring that screening was done by checking for results. The more aware that families and mothers are of the importance of the newborn screening program, the more likely they are to ensure that screening takes place.

The emergency newborn screening contingency planning efforts of the APHL identified several key elements that each state should integrate into a contingency plan. These include:

- Stocking of reserve testing reagents and supplies by laboratories and manufacturers;
- Backup plans to ensure diagnosis and follow-up services for infants identified in programs;
- Interstate and regional agreements through which back-up of laboratory capacity is ensured;
- Increased harmonization of laboratory disease panels and methods so that results are compatible among states;
- Availability of data systems to ensure record integrity and timely transmission of test results to providers and state programs.

A comprehensive report of Guidelines for the Public Health Laboratory's Continuity of Operations Plan (COOP) has been developed by the APHL (http://www.aphl.org/aphlprograms/ep/Documents/PHL_COOP_Guidelines.pdf).

More specifically, it becomes the responsibility of each newborn screening program to implement their COOP program and to notify hospitals and the public of changes through public service announcements and alerts. Communication plans are central to accomplishing this during emergency situations. The newborn screening laboratory should have identified backup laboratory capacity that is able to provide testing as mandated by the state in need or be positioned to inform families that screening panels have changed due to the emergency. Recent efforts by the Advisory Committee on Heritable Disorders of Newborns and Children to increase the uniformity of newborn screening panels should improve the ability of states to find appropriate back-up. Because of the potential impact of the disaster on laboratory operations, screening laboratories should have clear plans for the triaging of specimen testing to ensure that those positive screens associated with conditions for which medical emergencies can occur are prioritized.

In the event that specimen collection is impacted by an emergency, state programs should be positioned with contingency plans in place for obtaining specimens, testing the specimens, and communicating those results to patients and providers.

Because of various authorities available to a state, preparedness planning has been best organized in the systems over which the state has the most control, those being the screening laboratory and, to some extent, its referral network. However, a disaster can impact any critical component of the system beyond the screening laboratory including the availability of confirmatory clinical and laboratory diagnostics facilities and the functioning of manufacturers and shippers of laboratory reagents and time-critical therapeutics.

There are currently a number of national programs available or in development that can improve access to available assistance for newborn screening laboratories. The wide variability among states in the conditions mandated by the legislature of each state and offered in newborn screening is gradually subsiding as a stronger federal role is developing. Recommendations at the national level from the Advisory Committee on

Heritable Disorders in Newborns and Children are leading to greater similarity among programs and, therefore the ability of an external laboratory to assume responsibility for the panel of conditions mandated by that state. Programs such as the Laboratory Performance Program of the Region 4 RC are aligning the case definition of a screen positive infant. This simplifies the cascade of events that take place after the identification of a screen-positive infant. More consistency can be gained around other interstate variables including specimen storage rules and reporting requirements through similar national programs.

Diagnostic confirmation phase

Screen positive infants move into the diagnostic sector which, depending on the practices within their state, may operate under business agreements with the newborn screening program or within the private medical system. For the most part, the state's role in managing patients identified by newborn screening programs loosens as one transitions from short- to long-term management. This phase of the newborn screening program involves: (1) informing the appropriate providers of the results of the newborn screen; (2) communicating the results to the family, and tracking them down if displaced, if they were not among those initially informed; (3) promptly assessing the newborn's status and directing them into emergency care, if needed; and (4) evaluating the infant and seeking the appropriate testing to confirm the screening results.[7] For the most part, the state's role in managing patients identified by newborn screening programs loosens with the transition from short- to long-term management.

During the immediate aftermath of an emergency situation, after external resources have been able to access the disaster site, means of accessing highly specialized care or providers can be important. The NDMS can choose to involve appropriate clinical providers on their medical teams or develop mechanisms through which they can be accessed for consultation within the disaster site. Although disaster response organizations typically will have addressed their own communications needs, this does not necessarily extend to the local providers who remain within the disaster site, which is discussed below.

Managing a screen-positive infant's needs for immediate care during an emergency situation that has impacted the state's program and its ability to communicate is complex. Families and providers typically assume that after the collection of the specimen, no news is good news. It seems likely that the only way this component of the program can be protected is through the availability of electronic or telephone access that allows families to see that results are available or that allows providers to check for these results when seeing infants for the first time. Once informed, families may require assistance with transportation. The screening programs themselves require access to providers in different locations to improve the likelihood of finding a provider not impacted by the disaster.

An emergency may also have impacted the availability of, and access to, diagnosticians and clinical laboratories either local to the emergency or in other parts of the country. The more esoteric the clinical evaluation and laboratory testing that is required, the less likely it is that primary care providers can independently manage the situation. Metabolic disease and clinical genetics physicians are commonly involved in the delivery of highly specialized care related to most conditions in newborn screening. Pediatric hematologists and endocrinologists are similarly involved in diagnosis and management of related disorders. A wide range of backup plans can be developed by the various participants in the newborn screening program to ensure the availability of and access to appropriate clinical and laboratory providers. Directories that list the providers of laboratory and clinical services for these patients are in development in both the RCs and the NCC/ACMG and should be publicly available soon. A directory listing clinical laboratories performing specialized biochemical genetic testing is already available at http://biochemgen.ucsd.edu/UCSDW3BG/. Providers would be well served by establishing contact with potential backup laboratories and providers to ensure that they have the capacity to absorb the increased workload and by considering the full range of laboratory and clinical services that may be required for diseases in their programs.

Management and treatment phase (whether patients are identified by newborn screening programs or through clinical diagnosis)

Once diagnosed, some patients identified with genetic diseases may be placed on therapeutics whose continuation is critical, whereas the clinical needs of others may offer considerable flexibility. Treatments may entail medical procedures that may be highly specialized and not available in community hospitals, such as renal dialysis. In the effort to maintain a patient's therapeutic program, patients and families, manufacturers of the therapeutics, pharmacies, local institutions, providers, and agencies charged with mitigating the impact of an emergency on the public may need to become involved.

The first critical need for the diagnosed patient is to have access to their medical history and health records. In the absence of a national health information system in the United States, alternatives are available. Patients can be given key parts of their medical record and supporting information about emergency management in either digital or paper format. As with the Region 4 project with Midwest Emergency Medical Services for Children Information System, patients can also be given control over access to a centralized medical record and information, which they can allow providers to access as needed. Similarly, a number of private sector companies provide systems for storage of medical records, as through Microsoft's Health Vault™, Google Health™, Life Sensor HIPAA (Health Insurance Portability and Accountability Act) compliant online database, the Rubicon eHealth Manager™ system, and others. Simple storage devices such as flash drives or other memory devices that store information can be retained by consumers and used as needed. Providers and institutions can also take steps to ensure that backup copies of medical records are available if they cannot access those stored on-site.

Orphan therapeutics

Medical foods and orphan products are sometimes difficult to obtain even under normal circumstances. Suppliers contacted after Katrina were uniformly anxious to help, cutting through red tape and ensuring un-interrupted supplies of various metabolic formulas. Nevertheless, a pre-existing plan with a prepositioned, rotating stockpile of these items, in a central location, with dedicated courier service to ensure delivery where needed would mitigate some of the problems encountered in the Katrina aftermath. These supplies would include metabolic formulas and treatments for urea cycle disorders (Buphenyl®, Ammonul®, arginine, and citrulline) organic acidemias and aminoacidopathies (carnitine, B12, biotin, tetrahydrobiopterin, Lophenylax, Kuvan), and Cystagon to treat cystinosis. Identifying dedicated pharmacies in the affected area to provide products for neighboring regions would facilitate rapid access to rare and occasionally expensive medications.

Patients on treatment regimens must generally continue these during emergencies. Further, many of the therapeutics can be exceedingly expensive with limits imposed by insurance payers of access to only a 1-month supply at a time, limiting the ability to keep a backup supply. For potential disasters of a seasonal nature such as hurricanes, families may want to negotiate an increased supply of medical foods and drugs with medical suppliers during these periods. It is also possible to provide manufacturers with a prescription that allows them to direct ship their product to patients, if needed. Minimally, patients and families should retain contact information of the manufacturers of the therapeutics on which their child is being maintained.

Backup pharmacies are important to ensuring the availability of critical therapeutics at disaster sites. Areas with the potential for disasters that arise with little forewarning (e.g., earthquakes) should have backup pharmacies in areas away from the primary pharmacies as is available in Los Angeles County. The CDC is also a site that has the capacity to maintain a backup pharmacy of critical medications associated with newborn screening conditions. Because the CDC is often well integrated into the federal disaster response program, it can be a source of treatments available to NDMS teams and is well positioned to make them available to providers at disaster sites. Manufacturers may also be able to preposition critical therapeutics near disaster sites when possible.

POSTEMERGENCY

Mass casualty events

After Hurricane Katrina, over 80 genetics professionals traveled to Baton Rouge, Louisiana, to work at the Family Assistance Center where victim identification was carried out under the direction of the Louisiana State Police. They contributed significantly to the DNA identification of casualties. On completion, a web-based survey of the participants in the program that was conducted by the American Society of Human Genetics showed that 97% would like to see the genetics community actively involved in further development of the role of geneticists in mass casualty events. Among the resources needed are:

- Education modules tailored to the needs of genetics professionals involved in kinship analysis;
- More formal relationships between the genetics and forensics communities;
- Formal integration of the genetics community into DMORTs multidisciplinary identification team;
- A registry of genetics professionals who are prepared to assist in bridging the gap between forensic science and grieving family members;
- A mechanism through which laboratorians with expertise in DNA methodologies can augment workforce needs of local forensics laboratories;
- A web site through which:
  - training/education modules can be made available;
  - updates about DNA identification issues can be provided to members.

Awareness of federal, state, and local officials would be raised and access to the genetics community could be gained if these tools and activities are put intro place.

Over-riding issues

Communication in an emergency situation

Communication is critical during emergency situations. State programs take the lead in raising public awareness. They may be trying to track specimens and coordinate referrals for diagnostic confirmation. Patients may be seeking services and provider groups may be attempting to reconnect among themselves and with other providers and their patients. Web sites at multiple locations with critical information about program plans and contact information for the various entities involved in emergency preparedness and the development of centralized communication hot lines can be useful. Among the options available for direct communication are:

- Cellular telephones, if operational;
- Voice-Over-Internet protocols such as Skype that allow phones and/or computers to communicate;
- Video-conferencing equipment that allow for communication and sharing of images and files as well the direct delivery of care via telehealth systems;
- Satellite-based communication systems that may also provide a valuable backup means of ensuring communication.

Regardless of the communication strategy chosen, mechanisms for maintaining their power source should be available.

Legal issues

Numerous legal issues have to be considered in emergency preparedness. The magnitude of the disaster can impact what might be managed centrally as through the authorities that arise when EMAC declares a disaster versus what might be managed locally. Memos of understanding with those involved in backup services, interstate compacts, and other agreements can cover issues such as medical licensing, rules covering return and storage of materials, malpractice, and liability of responders and other issues.

Financing

Financing of backup services is an additional area to consider in the development of emergency preparedness plans. Costs must be tracked, purchasing requirements met, invoicing systems agreed to, and mechanisms for payment for services established. Methods through which backup diagnostic and clinical service providers can be reimbursed also should be considered.

Pre-event planning and disaster plan exercises

Disaster plans are only as good as the preparation taken to use them. They should be reviewed periodically and, where possible, they should be drilled. Areas for improvement can be identified through exercises or experience gained through prior emergencies. Given the wide range of potential emergency situations and variability in forewarning, redundancy becomes increasingly important. Everyone with an interest in the programs, including patients, newborn screening laboratories and associated follow-up activities, providers and their institutions and emergency responders, must assume overlapping responsibility for the continuation of all aspects of the program.

As the focus on newborn screening programs continues to increase and as the CDC rolls out its Contingency Plan for the programs, much work will need to be done. The newborn screening laboratories and programs have made significant progress in preparedness for the screening and short-term follow-up of those identified in the programs. However, considerably more preparedness needs to be done in order that responders are able to address the broad needs of these patients over both screening, short- and long-term follow-up and management. The Regional Collaboratives will be convening again to address this preparedness.

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APPENDIX

Appendix 1

Patient/family emergency preparedness checklist

Emergency information form (paper or electronic) with:

- Demographic Information ☐
- Diagnosis ☐
- Medications and metabolic formulas ☐
- Emergency medical treatment protocol ☐
- Allergies ☐
- Immunization history ☐
- Special devices ☐
- Baseline examination parameters ☐
- Laboratory and radiology findings ☐
- Procedure history ☐
- Critical resources (e.g., oxygen, electricity for pumps or ventilators)
- Contact information for:
  - Primary care provider ☐
  - Specialty care providers ☐
  - Manufacturers of critical therapeutics/devices ☐
- Information to access personal electronic health records ☐
- Information on web sites with back-up contact information ☐

Appendix 2

Draft: emergency shut-down back-up plan template

I. Purpose of plan
II. Scope and applicability
  A. Implementation triggers
    1. Nature of problem
    2. Estimated length of shut-down time
    3. Plan implementation
  B. Responders
    1. Laboratory and follow-up staff in emergency shutdown
    2. Healthcare facilities submitting specimens
    3. Laboratory providing back-up services
III. Authorities and contacts
  A. MOU/MOA – signed agreement with lab providing back-up
    1. Scope of emergency support services/operational relationship
    2. Funding of back-up services
    3. Ownership of specimens
    4. Liability

B. Contacts
    1. State Public Health Laboratory director of affected laboratory
    2. Director of laboratory providing back-up service
    3. Executive director of the APHL
    4. Newborn screening staff of affected laboratory
    5. USPS or courier service used to ship specimen
    6. State hospital association
    7. Vendors of reagents and kits

IV. Concepts of operation and roles and responsibilities
A. Implementation notification
    1. Public health laboratory director
    2. Back-up laboratory contacts
    3. Local emergency management agency (if applicable)
    4. APHL
    5. Staff of affected laboratory and follow-up coordinator
    6. Health care facilities affected by shut-down
    7. Specimen couriers
    8. Kit/reagent vendors
B. Modes of communication
    1. Phone
    2. Wireless
    3. Trunked communication system – computer-controlled radio system
    4. Alternate communication systems
C. Transportation and delivery of specimens
    1. Triage prioritized specimens or analytes (if applicable)
    2. Specimen tracking procedure
    3. Mode of transportation
        a. USPS
        b. Commercial courier
        c. Private conveyance
D. Reporting test results
    1. Roles and responsibilities of each party
        a. Screen positive
        b. Screen negative
        c. Responding to test result requests by submitters
    2. Mode of communication
    3. Turn-around-time expectations
    4. Data transfer
        a. Required data elements
        b. File formatting/translation program
        c. Secure data exchange requirements
        d. Database management
    5. Final report requirements (such as listing alternate test on report)
E. Specimen retention or return
F. Reimbursement procedure
    1. Tracking of costs or charges per specimen
    2. Purchasing requirements and codes
    3. Invoicing procedures

V. Post-emergency procedures
A. Set up phone hotline for parents
B. Review of specimen submission procedure
    1. All specimens submitted during shut-down were screened
    2. Procedure for testing specimens not prioritized during triage operation
    3. All screen-positive results have been followed up
C. Communication of return to pre shut-down activities

VI. Plan review and process improvement