NYMAC

NEW YORK-MID-ATLANTIC CONSORTIUM
FOR GENETIC AND NEWBORN SCREENING SERVICES

Delaware, District of Columbia, Maryland, New Jersey
New York, Pennsylvania, Virginia, West Virginia

Implementing the Affordable Care Act in the NYMAC Region

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SECTION 1: INTRODUCTION

The 2010 Patient Protection and Affordable Care Act (ACA) is a complex effort intended to expand the number of U.S. residents covered by health insurance, to reform health care financing and reimbursement, and to encourage innovation in health care delivery through a combination of regulatory reforms, incentive payment programs, new Federal agencies, and demonstration projects coordinated by the Department of Health and Human Services (HHS) and the Centers for Medicaid and Medicare Services (CMS). The law requires extensive monitoring of health outcomes to inform quality improvement over time, supported by improved and expanded use of health information technology [1].

Given its ambitious scope, there is optimism that the ACA can improve access to quality care for families with heritable disorders. Implementation of the ACA is currently underway. While some provisions are already in practice, others are far from being implemented. The full impact of the ACA on quality of care, outcomes and cost effectiveness has yet to be determined.

NYMAC, as an eight-member regional collaborative (seven states plus the District of Columbia) addressing the needs of persons with heritable diseases, is interested in the implications of the various aspects of the law for its populations. In the United States, a rare disease is one that affects fewer than 200,000 individuals. The National Institutes of Health (NIH) estimates that there are approximately 6,800 such conditions [2]. Most rare diseases are genetic (here, we use the term interchangeably with "heritable") and often impose severe and chronic disabilities affecting multiple organ systems, hampering quality of life, and truncating life expectancy in affected individuals. The economic, social and emotional burdens of caring for a family member with such a condition are profound [3].

The term "heritable disorders" refers to a diverse group of conditions that are not amenable to generalized description. Despite their heterogeneity, they raise four common types of challenges for affected families and health care providers by virtue of their rarity and severity [4]. First, because epidemiological data on rare diseases are so

difficult to obtain, standards of care for families with heritable disorders vary widely from provider to provider. This gives rise to inequity and confusion about the "medical necessity" of certain items and services. Where disease registries exist, they are often siloed, proprietary, and/or incomplete.

Second, as conditions in this group are generally not curable, they often require treatment for the duration of an affected person's life. Although the Orphan Drug Act of 1983 has incentivized the development and commercialization of therapeutics for rare diseases, there is evidence that the market exclusivity for so-called "orphan drugs" has led to unacceptably high, lifelong drug costs [5].

Third, timely diagnosis of a condition (and, when desired, prevention of affected births) relies on access to diagnostic testing, genetic counseling and reproductive health services. These services are not widely available or universally covered by health insurance.

Fourth, optimal care for individuals with heritable disorders often requires close coordination between many medical and non-medical providers. When a heritable condition is diagnosed, increased monitoring and multiple specialist referrals may be indicated, not only for an affected individual but for family members as well [6] [7] [8].

These challenges warrant careful consideration during the implementation of health reform at the state level, where there is ample room for further interpretation of the ACA's core principles. Despite this complexity, the ACA holds some promise for families with heritable conditions in the U.S. Some aspects of the ACA will almost certainly benefit this population, like the prohibition of coverage-denial based on pre-existing conditions, the removal of annual and lifetime dollar limits on benefits, and the ability of young persons to remain on their parents' health plans until age 26.

To complement these and other reform measures, patient advocates in the New York – Mid-Atlantic area must identify locales where coverage and services may remain inadequate or disparate for families with heritable conditions. Doing so will enable the formulation of targeted and well-informed advocacy on their behalf.

This review paper is intended to provide background information for NYMAC stakeholders and to support ongoing discussion of potential strategies and actions that can help issues in the region. It will be circulated for additional comments and input to guide efforts of NYMAC including the establishment of a NYMAC ACA workgroup to support future efforts and continued surveillance as the dynamics of implementation unfold.

This paper addresses implementation in two overall areas: the financial and delivery related aspects of the ACA. The information is examined to see how these factors might impact persons with heritable disorders at the national level. As many aspects of reform will be state-specific, the paper also explores some of the state-level implementation issues by providing some details of ACA implementation experienced in Maryland. A companion matrix provides a point in time overview of various aspects of ACA implementation and related issues in each of the NYMAC states, revealing a "bird's-eye view" of differences in approaches to and stages of implementation across the region.

SECTION 2: FINANCING AND REIMBURSEMENT REFORM

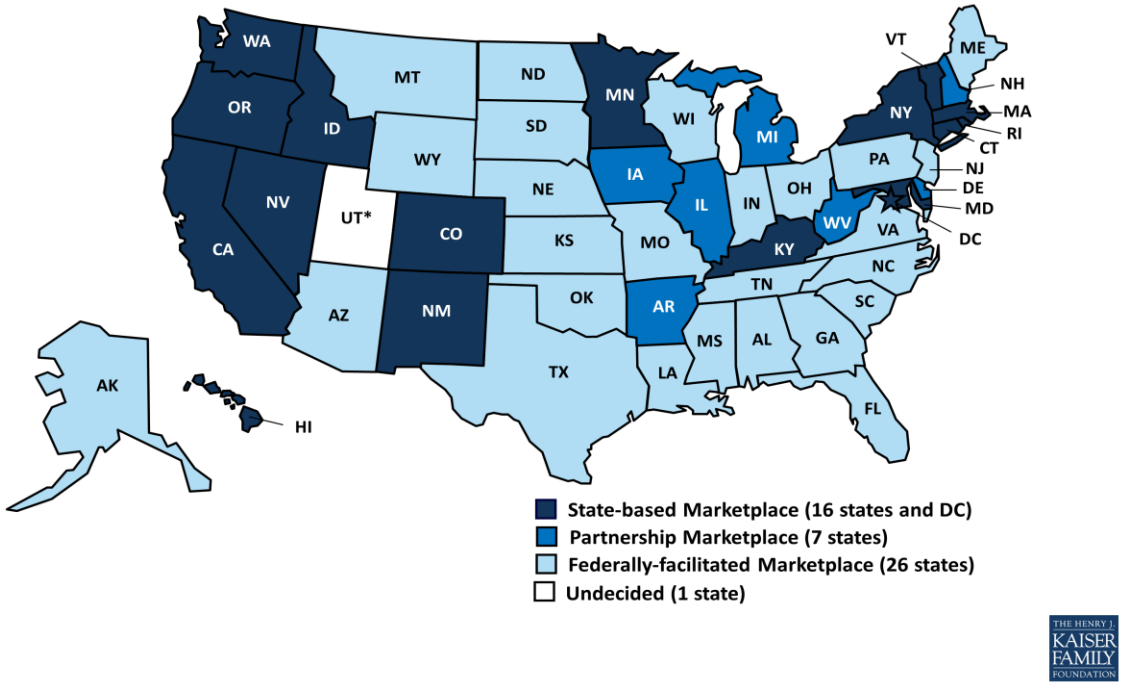
The Affordable Care Act sets up a new approach to insure most citizens by establishing Health Insurance Exchanges or Marketplaces that will operate in each state. These Marketplaces will select and offer plans, provide assistance to those shopping for a plan, and identifying the subsidies available to them. Plans sold in these Marketplaces will be required to cover all items on a list of essential health benefits (EHBs) stipulated in the ACA. Expansions in coverage through Medicaid are available, with major federal cost sharing, but states are not required to expand their Medicaid programs. Marketplaces are only likely to impact a small percent of the population. Most people will continue to get their coverage through public benefit programs (Medicaid/Medicare/CHIP) and groups that may be overlapping that include large group plans, self-funded plans, grandfathered plans or

A. Health Insurance Marketplaces

i. Selection of Marketplace Models

The structure for overseeing the insurance related provisions, including the essential health benefits and plan selection process is the Health Insurance Marketplace, (Marketplace) required by the ACA. The Marketplace will facilitate the processes by which individuals and small businesses can shop for affordable health coverage. States may operate their own exchanges or partner with the Federal government to do so. States choosing neither option must default to a Federally-run exchange. All exchanges were originally expected to be ready to open for enrollment on October 1, 2013 for coverage effective January 1, 2014. As noted on the map below, DC, New York and Maryland will operate their own exchanges with the rest of the eight states doing opting for a partnership model with the Federal government or a Federally Facilitated Marketplace.

States Decisions For Creating Health Insurance Marketplaces



Source: Kaiser Family Foundation: Establishing Health Insurance Marketplaces: An Overview of State Efforts May 02, 2013

ii. Aspects of Health Insurance Marketplace Implementation

The Marketplace model is intended to simplify the process of shopping for health insurance coverage while providing supportive services to healthcare consumers and heightening transparency. The ACA provides a variety of approaches to assist consumers

with making choices. These approaches might include: on-line assistance including web chat assistance, toll free call centers, brokers and in-person assistors, navigators and certified application counselors. The current companion matrix identifies some of these options in each of the NYMAC states as of that point in time. Options and their implementation are currently being developed including those for the federally facilitated marketplaces.

One of the options is the development of "navigators" to aid individuals in evaluating the relative merits and drawbacks of available policies and guide consumers to a plan that meets their needs. As Marketplaces begin to take shape, diverse interest groups have proposed requirements and restrictions on the kinds of information and assistance navigators will be allowed to provide consumers, politicizing their role in the implementation of reform [9]. While state-based Marketplaces must cover the cost of navigators through funding provided in part by the federal government, CMS is providing an additional $54 million through a competitive process for states with federally facilitated and state partnership market places. Entities or individuals serving as Exchange Navigators must have expertise in eligibility and enrollment rules and procedures, the range of qualified health plans and insurance affordability programs, the needs of underserved and vulnerable populations (such as rural populations and individuals with limited English proficiency), and privacy and security standards.

Families affected by heritable conditions need certain items and services in quantities and for durations that differ from what is required by the average health care consumer. To make sure they benefit from the establishment of Marketplaces, it will be important for Marketplace navigators to be familiar with their needs and interests in order to select an appropriate plan. This will include a better understanding of the details of coverage by a given plan.

While the navigators and other options to assist consumers can provide guidance and help in selecting plans, for persons with various heritable disorders, detailed information may not be available and consumers may need to directly call plans to identify whether specific benefits that they are interested in are provided for in that plan.

B. Essential Health Benefits

i. Overview

Beginning on January 1, 2014, the Affordable Care Act (ACA) will require all new individual and small group health plans to provide coverage of Essential Health Benefits (EHBs). Section 1302(b)(1) of the ACA specifies that EHBs must include items and services in the following 10 categories:

- Ambulatory patient services
- Emergency services
- Hospitalization
- Maternity and newborn care
- Mental health and substance abuse
- Prescription drugs
- Rehabilitative and habilitative services and devices
- Laboratory services
- Preventive, wellness and chronic disease management services
- Pediatric services including oral and vision care

The ACA's prohibition on lifetime and annual limits apply to all plans and policies with the exception of grandfathered individual policies.

HHS has instructed each state to choose an existing health insurance plan as a benchmark [10]. States are given four options for their starting benchmark plan:

1. The largest plan by enrollment in any of the three largest small-group insurance products in the state's small group market;
2. Any of the largest three state employee health benefit plans by enrollment;
3. Any of the largest three national Federal Employees Health Benefits Program plan options by enrollment; or
4. The largest insured commercial non-Medicaid Health Maintenance Organization (HMO) operating in the state.

In the NYMAC region, all states have benchmark plans as of March 2013.

ii. Issues for Families with Heritable Disorders

Coverage issues including essential health benefits and particular services included in benefit plans have specific relevance to families with heritable disorders. The HHS bulletin leaves room for states to vary the definition of an acceptable EHB package by selecting among one of ten plans. However, they were restricted from making adjustments in terms of benefits or limitations to the selected plan, other than bringing it up to ACA standards. The decision to implement the EHB rule this way was informed by reports of the Department of Labor (DOL), Mercer, LLC, the Kaiser Family Foundation/Health Research & Educational Trust, and the Institute of Medicine (IOM). They were supplemented with internal reviews of state and Federal employee plans. Together, these evidence reviews revealed three sources of variation in health coverage that may continue to impact families with heritable disorders in the U.S. under the ACA.

Findings from the review of existing plans found several variations across states that have potential implications for individuals with heritable disorders including cost-sharing provisions, inconsistent coverage of habilitative services, and variations in state benefits and mandates. These are elaborated below.

First, the reviewers found that existing plans differ more in their cost-sharing provisions than in the mix of items and services they cover [10]. This is significant because the ACA only specifies a list of benefits that must be covered by all new individual and small group plans. The terms of cost-sharing, including co-payment and deductible amounts, will be determined separately based on the actuarial value of each plan (i.e. “Platinum,” “Gold,” “Silver” or “Bronze”). In practical terms, this means that plans may differ significantly in the amounts they will pay for covered items and services. This means that families with heritable conditions could continue to face high deductibles and copayments for the specialist visits, ambulatory care, and habilitative services on which they rely more heavily than the typical patient under the Exchange as they currently do.

A second relevant finding was that coverage for habilitative services differed substantially among the different plans reviewed [11] [12]. This lack of coverage pre-ACA is not surprising since few plans covered such services before 2014. The ACA uses the definition of "habilitative services" adopted by the National Association of Insurance Commissioners (NAIC), as follows:

> *"Health care services that help a person keep, learn or improve skills and functioning for daily living. Examples include therapy for a child who isn't walking or talking at the expected age. These services may include physical and occupational therapy, speech-language pathology and other services for people with disabilities in a variety of inpatient and/or outpatient settings."* [13]

The lack of specificity in the ACA's definition is concerning since there are many aspects of what might be contained in coverage. In addition to the therapies listed above, habilitation involves items such as durable medical equipment (DME), orthotics, prosthetics, low vision aids, hearing aids, and a bevy of augmentative communication devices. According to the HHS, states may either a) cover whatever level of habilitative services they believe is appropriate (which HHS will review in 2016); or b) cover habilitation services on par with *re*habilitation benefits.

Neither of these options is fully satisfactory from the standpoint of families affected by heritable disorders. When HHS evaluates the benchmark approach to EHB provision in 2016, there is no guarantee that it will recognize any state-mandated benefits introduced after the passage of the ACA as "essential." Even more important is the distinction between rehabilitative services and habilitative services. Although both are delivered by the same workforce and involve similar interventions and devices, their primary aims are distinct. The goal of *re*habilitation is to help an individual regain skills that were lost due to an accident or illness. In contrast, the primary aims of habilitative services are to learn skills that are generally acquired during the normal course of living, slow a loss of skills or to maintain abilities that would otherwise decline as a result of an injury or health condition.

Owing to their individual goals, patients receiving habilitative care may require a distinct mix of items and services for a different duration than would be appropriate for a rehabilitation patient. For this reason, it will be important for plans and policies offered

through the Marketplaces to assess limits on habilitative services separately from limits on rehabilitation benefits.

Insurance coverage for habilitative services is crucial to individuals with heritable, disabling conditions so they can achieve their maximum level of functioning and independence. Lacking coverage for these devices and services is a common reason why affected individuals are often forced to exit the private market (including leaving employment) in favor of publicly supported programs such as Medicaid. The delays in treatment they must endure as they transition from one plan to another can have profound consequences for their well-being, independence and longevity.

A third relevant finding was that state benefit and provider mandates ("state mandates" hereafter) vary widely in number, scope and substance. State mandates are especially important for families with heritable diseases as a policy tool for filling in 'gaps' in coverage for items and services that may not be sufficiently covered under a state's EHB benchmark plan [14]. In the NYMAC region, each state has chosen a small group plan as its benchmark. While state mandated benefits passed prior to December 31, 2011 must be included in small group plans, which are state-regulated, it remains unclear whether and how future state mandates will co-exist with ACA implementation plans.

The potential importance of state mandates is well illustrated by the inter-state variability of insurance coverage for medical nutritional therapy (MNT), on which individuals with inborn errors of metabolism (IBEM) rely. MNT is the most effective treatment for these disorders that can cause profound mental and physical dysfunction when untreated. Although the precise cost of supplying MNT to an individual with an IBEM varies depending on the condition in question, it is estimated that for one such condition, phenylketonuria (PKU), it costs $1900 - 2500 per year to feed a newborn (according to a licensed medical nutritionist) and up to $8,522 per year to feed late adolescent or adult males in 2010 [15]. The following provides some estimates of the range of estimated costs.

Age*	# Cans/Month*	Monthly Cost*	Annual Cost*	Nutricia NA Estimate**
Infant				$2,000
Child	4-10	$159-$397	$1,908-$4,764	$5,250-$7,750
Teen	8-10	$317-$397	$3,804-$4,764	$6,500
Adult	12	$476	$5,712	$8,500-$11,000

*Note: Based on $238 per case of six cans for Phenex-2 unflavored when purchased from the manufacturer. Type of formula varies based on IBEM disorder, age, and physician prescription. Also, this table does not reflect the price mark-up of formulas purchased through a pharmacy. Source: VCU Metabolic Treatment Center and Abbott Nutrition

** Nutricia North America consumer price list for metabolic formulas. Also see table 3 in the pdf from JLARC 2008 report annual PKU formula cost for child is 1908-4764$, p. 22

The FDA regulates MNT under its food and nutritional supplement statutes and not as a "drug." As a result, there is much inconsistency in the terms of insurance coverage for this essential therapy. Although 38 states now require MNT coverage, caps, limits to eligibility, age limits, or narrow definitions of what constitutes MNT limit the effect of these mandates.

The patchwork of state mandates has resulted in disparate access to MNT treatments across the country. The situation is paradoxical: while the ACA will require health insurers to pay for all newborn screening tests recommended by HHS, it does little to facilitate access to the life-saving treatments that renders such screening worthwhile from individual and public health standpoints.

Going forward, it will be important for advocates for patients with heritable disorders to identify where and how state mandates play a key role in determining access to items and services like MNT. This will help shape an agenda for reconciling these state mandates with regional ACA implementation plans in the future. A recent issue brief

provided a review of habilitative services coverage for children under the ACA. This review identifies a number of issues that are anticipated and a table of how the decisions are likely to be made based on the type of plan and current status of habilitative benefits in a given state. [16]

C. Medicaid Expansion

The ACA provides opportunities to expand Medicaid eligibility and enrollment. The new rules will take effect on January 1, 2014. In a case known as *National Federation of Independent Business vs. Sebelius* in 2012, the U.S. Supreme Court determined the ACA's Medicaid expansion to be unconstitutional on the grounds that it is unduly coercive because the Secretary of HHS could potentially withhold all of a State's Medicaid's funds as a penalty for non-compliance. As a result of this ruling Congress remains free to extend additional Medicaid funds to each state for the purpose of enacting an expansion. However, the Federal government cannot make funding a state's entire Medicaid program contingent upon its compliance with this aspect of the ACA. The Supreme Court ruling excluded children 6 to 18 who will be covered up to 138% of poverty.

State implementation of Medicaid expansion is of special concern to families affected by heritable conditions, many of which involve some degree of disability, because Medicaid is the primary payer for the long-term care services on which so many individuals with disabilities rely [17]. Even if the individual does not need long-term care, many affected adults cannot work full-time because of their condition. The parent(s) of an affected child might also be unable to work full-time because of the child's special needs. (See estimates in the companion matrix). As health insurance is typically provided as an employment benefit, these individuals also rely heavily on Medicaid.

Currently, criteria for Medicaid eligibility vary from one state to another. The companion matrix provides information on pre-ACA Medicaid eligibility by state. Federal law requires Medicaid to cover certain core groups of people, such as pregnant women and low-income parents and children, up to a minimum income threshold. States may extend coverage to members of these groups at higher income levels subject to a state-determined ceiling. Thus individuals with disabilities may currently qualify for

Medicaid based on federal and state criteria while in most states single adults without dependent children do not.

The ACA eases Medicaid eligibility criteria by raising the income threshold effectively to 138% of the Federal Poverty Level (FPL) given a 5% income disregard ($26,344 per year for a family of three in 2012). Although ACA also establishes a Modified Adjusted Gross Income (MAGI) methodology for determining financial eligibility, it exempts adults who qualify for Medicaid based on disability from having their eligibility assessed by this more stringent method [18].

While some Medicaid eligibility standards are mandatory for states that choose to participate in the Medicaid program, others are determined by the state. For example, some states offer Medicaid coverage to people who are in need because of high out-of-pocket medical expenses, even though their incomes exceed Medicaid eligibility limits.

Similar to the private benchmark plan, the Medicaid benchmark package will consist of benefits that must be provided to all newly eligible individuals after the expansion takes effect. The implications of the state expansions will have differential impacts on families with heritable conditions due to the eligibility criteria and benchmark benefits each state selects. In states where the increased income eligibility will be an expansion for children, these children will also get regular Medicaid including EPSDT.

D. Financing and Reimbursement Reform: A Summary

A state's choice of a benchmark plan will be significant for families with heritable conditions because it will specifically define which items and services are considered part of EHBs in their state. The 10 plans from which states could choose might not cover certain items and services comprehensively. For this reason, it will be crucial for NYMAC to identify circumstances and areas where benchmark plans are expected to fall short, so that policies for supplemental coverage can be proposed.

The ACA's Medicaid expansion holds some promise of extending coverage to a larger number of individuals with heritable disorders. However, its impact could vary

depending on the specific implementation plan chosen in each state. Existing state mandates in relation to ACA implementation are not currently at risk. A regional inventory of state mandates that benefit families with heritable conditions will help NYMAC to ensure they are either upheld or incorporated into state-level ACA implementation plans.

E. Maryland ACA Benefit and Health Insurance Marketplace Implementation Efforts

i. Legislative History

The Maryland Health Benefit Exchange Act of 2011 established the Maryland Health Benefit Exchange (MHBE) as a public corporation and independent unit of state government. Working with the Department of Health and Mental Hygiene, the Department of Human Resources, and the Maryland Insurance Administration, the MHBE is creating a marketplace, the Maryland Health Connection (MHC) where Maryland residents and small businesses will explore health insurance plans, compare rates, and determine their eligibility for tax credits, cost sharing reductions and public assistance programs such as Medicaid and the Maryland Children's Health Insurance Program [19]. At its inception, Governor Martin O'Malley created a nine-member board to oversee the planning and implementation of the exchange called the Health Care Reform Coordinating Council[20].

Currently, the MHC has four standing advisory committees overseeing operations in the following areas: continuity of care, plan management, finance and sustainability, and outreach via the exchange's Navigator program. The MHC also operates a steering committee which examines issues related to risk adjustment, reinsurance, and risk corridors [21].

The Maryland Health Progress Act of 2013 was signed into law on May 2, 2013. The bill updates Maryland's health policies to include revised insurance rules, new Medicaid eligibility thresholds, and more precise terms of operation for the MHC. Beginning in 2015, the legislation will require the cost of running the MHC to be covered by an existing premium tax on participating insurance companies.

ii. Maryland's Health Benefit Exchange (Marketplace)

Certification criteria for the MHC's Qualified Health Plans will be phased in over a period of two years following the start of MHC's operation in October of 2013. In the MHC's first year of operation, participating insurance carriers will be allowed to define their own network adequacy standards. They will be required to report quarterly data about the kind of providers they include in their networks, the accessibility of their services, and their members' patterns of service utilization. MHC will develop longer-term criteria for payer accreditation informed by data collected in during the MHC's inaugural year of service [21]. Qualifying health plans participating in the MHC will need to offer at least one silver-level plan and one gold-level plan outside the Exchange, with a limit of no more than four different benefit designs per level in the individual insurance market. The exchange will review all participating carriers annually. The MHBE has designed a multi-step process to authorize carriers and certify benefit designs for the MHC. The deadline to apply to become an MHC carrier was May 1, 2013.

a. Outreach Activities

In the first half of 2013, MHC focused heavily on consumer assistance and outreach. Inquiries about the exchange, insurance enrollment, and/or Medicaid eligibility will be fielded by a Consolidated Service Center, scheduled to begin operation in June. The MHC will also contract with one Connector Entity (CE) in each of six geographical regions, a role that may be filled by one single organization or a consortium of several. Each CE must employ certified Navigators, who will be supervised by one Exchange Navigator Program Manager per region. Distinct Navigator programs have been established for the small group and individual markets. CEs may also solicit assistance from additional, uncertified personnel to provide services including consumer education and application counseling [22]. A list of organizations serving as CEs in Maryland can be found in Appendix B.

b. Essential Health Benefit Benchmark Plan

The HCRCC selected the state's largest small group health plan by enrollment, CareFirst BlueCross BlueShield HSA Open Access, to serve as its benchmark plan. To compensate for the plans omission of pediatric dental and vision benefits, the HCRCC chose to supplement its benchmark coverage with the existing Maryland Children's Health Program benefit and Federal Employee Plan Blue Vision High plans. The HCRCC has also demanded that the plan's coverage for adult habilitative benefits match its existing coverage for rehabilitative services in individuals aged 19 and older. Mental health coverage will be supplemented with the Federal Employee's Health Benefit Plan.

Under Maryland's benchmark plan, quantitative limits exist on the following services: skilled nursing services, outpatient rehabilitation services, and diagnostic testing. Hearing aids for members over 18 years of age are not covered. Furthermore, the plan does not cover long-term custodial care or private-duty nursing. To qualify for unlimited nutritional therapy, medical foods, and pulmonary services, members must be diagnosed with a condition known to require such therapy and must receive care from a qualified practitioner [23].

iii. Rate Review in Maryland

The ACA requires the state or federal government to review proposed increases in health insurance premiums based on granular assessments of health care quality and insurers' financial sustainability. Under a so-called "80/20 rule" insurance companies must reveal how many premium dollars they spend on health care relative to other costs. And they must spend at least 80% of its premiums on medical care and quality, or return the difference to premium payers. Since these provisions of the ACA took effect in 2010, Maryland has received two federal grants to help build upon the longstanding work of Maryland's Health Services Cost Review Commission, a state agency that has held broad powers of hospital rate setting and public disclosure since 1977 [24]. Maryland's rate review commission is the only remaining such organization in all 50 U.S. states.

As part of its rate review enhancement efforts, the Maryland Insurance Administration hired an independent consultant to assess the strengths and weaknesses of its rate review program. Using a scale of 1 (weak) to 4 (strong) to assess the stringency

of Maryland's existing rate review process, the consulting agency gave Maryland an overall score of 3.5. Since 2010, the MHC has created several new positions to ensure dedicated attention to the continuing improvement of its rate review process as new data sources become available [25].

iv. Medicaid Expansion in Maryland

Maryland's State Medicaid program is moving forward with an expansion plan to include all adults with incomes under 138% of the FPL. As of January 2014, an estimated 20,000 Marylanders will be newly eligible for Medicaid. Maryland currently provides a limited-benefits package to approximately 88,000 low-income, childless adults under its Primary Adult Care (PAC) Plan. Starting in 2014, all PAC beneficiaries will automatically be enrolled into a full Medicaid plan. At present, the PAC plan operates under a 50/50 match rate. Since the federal government will cover the entire cost of Maryland's Medicaid expansion from 2014 to 2016 and will fund the program at a 90/10 match rate thereafter, transitioning PAC beneficiaries to Medicaid in 2014 makes fiscal sense (a projected $161 million in net savings in 2016) [26].

Also starting in January 2014, eligible Medicaid beneficiaries will be able to enroll using the MHBE and will receive eligibility determinations in real time via its website. In-person enrollment will also remain an option. Medicaid has hired a marketing firm to raise public awareness of the new Medicaid eligibility criteria and enrollment procedures. Enrollment support will also be available through the MHBE Navigator program. Two new MCOs have applied to be Medicaid providers, raising the number of Medicaid MCOs operating in Maryland to 10. Individuals applying to Medicaid for long-term care or under the Employed Individuals with Disabilities program will not face any changes in eligibility requirements and enrollment processes.

SECTION 3: SERVICE DELIVERY REFORM

A core aim of the ACA is to restructure the fragmented U.S. health care delivery system to favor patient-centered, coordinated care while slowing an unsustainable trend

of rising health care costs [1]. The ACA efforts build on earlier progress made as a result of the American Recovery and Reinvestment Act of 2009 and the Children's Health Insurance Program Reauthorization Act of 2009. It contains numerous measures to improve quality and efficiency while constraining health care spending. A variety of initiatives and demonstrations are included to address innovations in health care delivery including approaches involving new organizational structures, processes designed to change actual care delivery, financial incentives to support desired changes and technology and workforce support. As there is inadequate knowledge about the most effective approaches, CMS is funding these projects and assessing their potential wider applicability. Their current efforts primarily focus on Medicare beneficiaries but may also include other populations. A recent second round of grants will shift to address the Medicaid populations and are more likely to include pediatric populations.

States in the NYMAC region are involved in many of these efforts as summarized in the companion matrix. The following section outlines some of the key service delivery reforms underway. Many of these reforms are works in progress and while few involve pediatric populations, the findings from these efforts may in fact guide future policy decisions that impact our populations. Attention to these efforts should include examination of what their expansion would mean to families with heritable disorders and what might be required to better address their needs.

A. Developing New Organizational Models for Delivering Care

Among the new organizational models supported by the ACA are Accountable Care Organizations (ACOs) and Patient-Centered Medical Homes (PCMHs). The ACA also emphasizes the expansion of primary care and care coordination efforts. There are both explicit and implicit implications for developing and improving integrated health care delivery systems and reducing the current siloes.

i. Accountable Care Organizations (ACOs)

In November of 2011, CMS finalized a rule creating the new Medicare Shared Savings Program (MSSP) to reduce fragmentation and promote quality care for Medicare

beneficiaries [27]. The MSSP's main vehicles of reform are ACOs, voluntary associations of hospitals, doctors, and other health care entities who provide coordinated, high-quality care to Medicare beneficiaries while adhering to an aggregate spending limit defined by CMS each year. The incentive to remain within this spending limit arises from the opportunity for participating health care providers to share a proportion of the cost savings achieved after their first year of ACO membership. To earn shared savings, providers must fully and accurately report their performance using thirty-three quality metrics that cover four domains of activity: patient experience, care coordination and patient safety, preventive health, and caring for at-risk populations.

CMS is financing a number of demonstrations to develop ACOs. While mainly targeting Medicare and to some extent Medicaid beneficiaries, an ACO demonstration targeting Medicaid and the Children's Health Insurance programs was proposed but not funded. Four NYMAC states, Maryland, New Jersey, New York and Pennsylvania, are recipients of CMS ACO grants. In addition provider groups including hospitals and payers are initiating their own efforts, which often have a broader population focus. The companion matrix identifies NYMAC states with CMS ACO efforts. However, limited information is available on the growing development of other ACO efforts in the states at this time.

Skeptics of the ACO model point to prior failed attempts at implementing provider-led cost controls during the 1990s. One source of concern is that in conforming to ACO spending limits, certain providers may be hamstrung by large transition costs. This in turn may compel them to recoup their losses by forcing private insurers to pay higher rates, imposing higher cost-sharing requirements on enrollees with commercial insurance. Another concern is that under the ACA, ACO members will still be paid on a fee-for-service basis meaning that high-paid specialist practices will have little incentive to change the status quo. That said, given the novelty and variety of experimental delivery system reforms authorized by the ACA, it is premature to speculate about their full range of implications at this time.

ii. Patient-Centered Medical Homes (PCMHs) and Health Homes

The concept of a medical home, first introduced by the American Academy of Pediatrics (AAP) in 1967, was originally envisioned as an "accessible, family-centered, continuous, comprehensive, coordinated, compassionate, and culturally effective" model of care [28]. However, the PCMH has defied precise definition, hampering efforts to study its benefits and drawbacks systematically. The modern PCMH is reimagined as a home-base where a family can receive coordinated, holistic care through a dedicated case manager, access to multiple specialists, and evidence-based care that improves in response to data collection over time. Since 2011, the National Center for Quality Assurance (NCQA) has instituted a PCMH education and certification program in an effort to standardize and support the process of transitioning primary care practices to the PCMH model, also laying the groundwork for more systematic outcome measurement over time.

Under the ACA, the PCMH model is a viable way of meeting the health and other service needs of families affected by heritable and chronic conditions. By sharing responsibility for care coordination with families, and by facilitating links to other important social services in the community, a successful PCMH can reduce the emotional and financial burdens facing families with heritable disorders [29]. Several studies of families raising children with special needs have found that receiving care from a PCMH can reduce a family's risk of experiencing financial hardship, alleviate pressure to withdraw from the workforce, and reduce out-of-pocket health care expenses [30] [31].

At the state level some states are supporting PCMH initiatives including increased Medicaid payments based on the level of NCQA certification. While PCMHs emphasize primary care, a new effort relevant to persons with heritable disorders is looking at patient-centered specialty care. Relatively new and currently focused on major adult diseases, there is potential promise for specialty care that represents specialists who care for persons with heritable disorders although current early adopters under the CMS program do not include these specialists. Within the NYMAC region, there has been

considerable uptake in medical practices obtaining NCQA certification leading to a designation of being a leading medical home state for Maryland, New York, Pennsylvania and West Virginia. Details of state efforts and numbers of medical homes are included in the companion matrix.

In addition to encouraging medical homes, the ACA provides for the development of health homes, providing planning grants and technical assistance for states to develop special plan amendments (SPAs) to their Medicaid plan. The ACA made this option available as of January 2011 and provides for enhanced federal match of 90% for a limited period. The health home model of service delivery expands on the traditional medical home models and focuses on persons with multiple chronic conditions.

The approved services under the SPA are intended to expand care coordination, the integration of medical and behavioral health and building additional linkages for the system of care. The scope of services that can be reimbursed include those related to these three areas and encompass transitional care from inpatient to other settings that include follow-up; individual and family support and referrals to community and social support services. Provider arrangements that can provide a health home include providers such as physicians or physician practices, health centers and mental health centers. Teams are expected to be interdisciplinary. While a number of NYMAC states have submitted plans for health homes, only New York is currently approved for individuals with chronic behavioral and mental health conditions.

B. Additional Efforts to Address Primary Care and Care Coordination

Underlying the organizational models provided by ACOs, PCMH and health homes are the importance of both primary care and the need to incorporate approaches to coordinate patient care. The issue of care coordination for children with special health care needs has long been an important feature of federally supported programs targeting this population. ACA initiatives further emphasize these important processes including using NCQA care coordination standards. These standards include test tracking and follow-up, along with related lab practices, specifically naming NBS; referral tracking

and follow-up; and coordination with other types of facilities and providers, including non-medical providers. Further emphasis is placed on team approaches to ensure all patient needs are addressed including medical and support services. ACA efforts include both demonstrations and financial incentives.

i. Demonstration Projects Supporting Primary Care

The CMS Center for Innovation is funding some specific efforts addressing primary care. One demonstration, the Federally Qualified Health Care (FQHC) Advanced Primary Care Demonstration project, is a 3-year pilot initiative designed to evaluate the effect of the PCMH, sometimes referred to as the "advanced primary care" model. A joint collaboration between CMS and the Health Resources and Services Administration (HRSA), the project's goal is to determine if the PCMH model can improve care, promote health outcomes, and reduce health care costs among Medicare beneficiaries. Participating providers must commit to obtaining Level 3 recognition from the NCQA in return for monthly subsidies intended to assist with the transition costs involved in establishing a PCMH. The participants must also engage in care coordination practices that are recognized by NCQA. The program has enrolled approximately 500 primary care practices nationwide since November of 2011 [32]. All NYMAC states with the exception of Delaware have CMS demonstration projects related to primary care.

Another initiative, the Comprehensive Primary Care Initiative (CPCI) is a limited demonstration project occurring in seven states, in which Medicaid is working with commercial and state health insurance plans to allocate bonus payments among primary care practices that demonstrate improved care coordination. Within the NYMAC region, there is one statewide initiative operating in New Jersey and a regional initiative in New York. Under the program, monthly fees are awarded to primary care providers who help chronic disease patients follow through with personalized care plans, offer 24-hour services, and deliver preventive services. Since August of 2012, 497 primary care practices have enrolled in the program nationwide [33].

A third effort, operated at the state level, is the Multipayer Advanced Primary Care demonstration. New York and Pennsylvania are participating in this demonstration, which provides a care management fee for patients in Advanced Primary Care practices. Multiple payers including Medicaid and private plans are involved. Participating states are also expected to coordinate these efforts with the state's health promotion and disease prevention efforts.

Finally, a more targeted effort called Strong Start for Mothers and Newborns, which is jointly run by CMS and HRSA, is designed to address preterm births and improve outcomes for pregnant women and newborns. Efforts will test new approaches to prenatal care by various provider groups. NYMAC states are major participants in this initiative with the exception of Delaware and New York. More detailed information is provided in the companion matrix.

ii. Financial Incentives to Support Desired Changes

Various financial incentives are being developed and implemented to promote the growth of primary care practices, to encourage care coordination and award payment based on outcomes and episodes of care. These efforts target the delivery of and payment for care thus encouraging care coordination, and increased efficiency and quality. These demonstrations include bundled payments, value-based purchasing plans, and payment alignment for Medicare and Medicaid. While some of these efforts may not specifically address families with heritable disorders, they are included to describe current efforts to change how payments are being designed to influence delivery system reforms. These potential changes may have limited implications at this point, but if adopted broadly and/or expanded in terms of target components of the system and specific diagnoses and procedures, need to be kept in mind.

a. Bundled Payments for Care Improvement (BPCI)

To complement the ACO's emphasis on cost control, CMS has also introduced four versions of a "bundled" payment model designed to provide financial incentives to inpatient and post-acute care coordination for Medicare beneficiaries. This system addresses the fact that more than 75% of Medicare spending is devoted to patients with

five or more chronic health conditions who currently receive little by way of transitional care, comprehensive medication management, health coaching or navigation support[34].

Under the BPCI initiative, there is a list of 48 "episodes of care" for which CMS will reimburse at rates lower than the existing fee-for-service amounts. The defined episodes of care include nutritional and metabolic disorders, which directly address the NYMAC population as well as several others that may have some relevancy. The program allows several ways to reconcile actual expenditures against CMS target prices for these "episodes of care." Despite the long list of care episodes covered by the BPCI, providers retain considerable control over which conditions to bundle and how to allocate payments among participating providers within an ACO [35]. Four NYMAC states, New Jersey, New York, Pennsylvania and Virginia have multiple grants.

b. Value-Based Purchasing Plans

Since the enactment of the ACA, HHS has authorized a number of programs that link payment rates directly to the quality of care delivered by Medicare providers. Providers are rewarded or penalized based on their performance according to specific quality measures and indicators of utilization including hospital readmission rates and incidence of hospital-acquired infections. Separate value-based purchasing plans have been established for hospitals, physician practice groups, and skilled nursing facilities [36]. Whether these efforts might expand beyond Medicare is not known.

c. Financial Alignment of Medicare and Medicaid

Individuals eligible for both Medicare and Medicaid ("dual eligibles") are among the sickest and poorest citizens covered by federal insurance programs [37]. In 2010, there were an estimated 9.9 million such beneficiaries consisting mainly of low-income seniors or individuals with disabilities. Medicare tends to cover prescription drug, acute care and hospital bills for this population, while Medicaid supplements their Medicare premiums and long-term care costs. Although dual eligibles experience some of the most complex chronic conditions requiring coordinated care, inconsistent consumer protection

and eligibility requirements across Medicaid and Medicare programs have made it challenging to meet their needs [38].

To improve care coordination for dual eligibles, CMS has established a Federal Co-ordinated Health Care Office (FCHCO) in 2010. In 2012, the agency proposed two novel payment models intended to facilitate the financial alignment of Medicare and Medicaid for beneficiaries of both programs, one is capitation-based and the other is a managed fee-for-service model. To date, CMS has received financial alignment proposals from 26 states and has offered planning assistance to Massachusetts and Washington, which will each pilot one of the new payment models beginning in during 2013.

D. Opportunities to Foster Patient-Centered, Quality Care

i. Expanded Newborn Screening

Under the Affordable Care Act (ACA), parents may request newborn screening tests for conditions not typically screened for by their state, which must be covered by insurance. This is to ensure that their babies are screened for all conditions on the Recommended Uniform Screening Panel (RUSP) recommended by the Secretary's Discretionary Advisory Committee on Heritable Diseases in Newborns and Children (DACHDNC). Insurance companies have one year from when a condition is added to the RUSP to comply with the ACA's newborn screening mandate. At the moment, no state screens for every condition on the RUSP.

Targeting individuals with conditions identified through newborn screening is a unique opportunity for quality improvement. The Maternal and Child Health Bureau of HRSA funds seven Regional Collaboratives for Genetic and Newborn Screening Services, which cover all states and territories in the U.S., as well as a National Coordinating Center (NCC) to improve health-care delivery. A working group of the NCC has recommended that key activities to improve follow-up care will include explicit care coordination, clearly designated management structures, and equal partnerships involving affected families, primary care providers and specialists [39].

ii. Focus on Patient-Centered Outcomes

Since 2012, the Hospital Consumer Assessment of Health care Providers and Systems (HCAHPS) is being used to calculate value-based incentive payments to hospitals. HCAHPS is a measure that uses information about the patient experience to assess physicians and health care facilities, including nursing homes. The ACA established the Patient-Centered Outcomes Research Institute (PCORI) to identify research projects that provide quality, relevant evidence on how diseases and health conditions can be effectively diagnosed, prevented, treated and managed [40]. The objective of funded research is to provide information that supports people and their caregivers to communicate and make informed decisions. The priority areas include assessment of prevention, diagnosis, and treatment options.

For individuals with heritable disorders, the development of patient-centered and disease-specific outcomes will be essential if families are to benefit from the sweeping service delivery changes implemented under the ACA. Procedures and treatment regimes that would seem excessively costly for an average member of the population may be essential to someone with a heritable disorder. For example, while yearly mammograms are recommended starting at age 40 and continuing for as long as a woman is in good health, women with Neurofibromatosis type 1 may require surveillance via breast MRI at younger ages owing to their increased risk of malignancy and the difficulty of distinguishing malignancies from the disorder's characteristic neurofibromas. However, with the current dearth of evidence and relevant outcome data, the health care system may not always be attentive to these needs.

Patient-centered outcomes may also help to capture the value of health care products and services whose value is not measurable using existing means. For example, for some families, undergoing genetic testing may shorten a long diagnostic odyssey and obviate the need for a child to undergo a series of invasive and expensive procedures. Although arriving at a diagnosis this way may not change the affected individual's long-term medical prognosis, it enables a family to divert its emotional energies and financial resources to caring for a loved one in a more targeted fashion. It may also enable a family to screen other individuals at risk, explore reproductive options to prevent the

recurrence of a condition, and begin the process of psychological and emotional adaptation to a loved one's diagnosis. Existing outcome measures, do not reflect these sources of value in genetic diagnostic testing.

iii. Efforts to Make Health Care Safer

The Centers for Disease Control and Prevention (CDC) estimate that each year at least 1.7 million health care-associated infections occur in the U.S. causing 99,000 deaths. In addition, adverse medication events cause more than 770,000 injuries and deaths each year, and the cost of treating patients who are harmed by these events is estimated to be as high as $5 billion annually [41, 42].

The ACA includes a number of measures aimed at reducing unnecessary hospital readmissions and the risks of injury and infection from care. It authorizes the creation of Patient Safety Organizations (PSOs) charged with improving the quality and safety of health care delivery. Organizations that are eligible to become PSOs include public or private entities, profit or not-for-profit entities, provider entities such as hospital chains, and other entities that establish special components to serve as PSOs. PSOs create a secure environment where clinicians and health care organizations can collect aggregate and analyze data, thereby improving quality by identifying and reducing the risks and hazards associated with patient care. In addition, Partnership for Patients, a national public–private initiative, has set a goal of reducing unnecessary hospital readmissions by 20% from 2010 to 2013.

For families with heritable disorders, the issue of hospital-acquired injury is often overlooked. However, many routine treatments may be contraindicated for individuals with rare, heritable conditions. For example, common medications such as corticosteroids, valproic acid, phenytoin, barbiturates and propofol can be toxic to patients with certain mitochondrial disorders [43]. A transportable electronic medical record will allow such contraindications to be clearly reflected in a patient's electronic health record (EHR) during routine and emergent situations.

D. Supporting Changes Through Technology and the Workforce

Changes in the service delivery system have implications for the development and use of information technology and for the workforce that will be needed. Both are recognized in the ACA although the building of the health information technology (HIT) infrastructure began earlier than the ACA. Workforce issues have been the focus of federal efforts for many years but the ACA recognizes some new demands and expectations.

i. Improving Health Information Technology Infrastructure

The 2009 Health Information Technology for Economic and Clinical Health (HITECH) Act established programs to improve health care quality, efficiency, and safety through the adoption of HIT including (EHRs). Under the HITECH Act, eligible health professionals and hospitals can qualify for Medicare and Medicaid incentive payments when they adopt certified EHR technology and use it to achieve specific stages of meaningful use (MU). A description of the different phases of MU can be found in Table 1.

Table 1: Stages of Meaningful Use

Stage 1: Data Capture and Sharing (2011-2012)	**Stage 2: Advance Clinical Processes (2014)**	**Stage 3: TBA (2016)**
Electronically capturing health information in a standardized format.	More rigorous health information exchange.	Improving quality, safety, and efficiency leading to improved health outcomes.
Using that information to track key clinical conditions.	Increased requirements for e-prescribing and incorporating lab results.	Decision support for national high priority conditions.
Communicating that information for care co-ordination purposes.	Electronic transmission of patient care summaries across multiple settings.	Patient access to self-management tools.
Initiating the reporting of clinical quality measures and public health information.	More patient-controlled data.	Access to comprehensive patient data through patient-centered Marketplace.
Using information to engage patients and their families in their care.		Improving population health.

Source: www.healthit.gov

To date, four series of regulations have been issued pursuant to the HITECH Act. Two of these, issued by CMS, define sequential phases of MU. The other two, issued by the Office of the National Coordinator for Health Information Technology (ONC), identify certification criteria for EHR technology.

The increased use of EHRs has significant potential to benefit families affected by heritable disorders by making it easier for their caretakers to communicate, coordinate, and track the many facets of their complicated care. Beyond their role in facilitating care coordination, EHRs will be important to support the increased use of next-generation sequencing and genome-wide testing in health care. This is because detailed phenotype description, family history information, and lab results are crucial to gleaning insight from the results of genomic testing. Documentation of this information presents challenges because many of the symptoms affecting individuals with heritable disorders are variable and non-specific.

One system, originally developed for use in a research context, demonstrates the potential of electronic recordkeeping to help integrate a genetics and dysmorphology vocabulary into everyday practice. PhenoDB, a tool under development at Johns Hopkins University, now includes an ontology of over 10,000 standardized terms derived from entries in the Online Mendelian Inheritance in Man Database (OMIM), which are linked to an online record system supporting the activities of the Centers for Mendelian Genomics (CMG) [44]. Although the CMG was developed as a research project, many of the tools used to capture, standardize and link clinical data to lab data may have applications in clinical care as well. As such, they should be of interest to those who wish to see better integration of genetics terminology and family history data into the everyday workflow of health care providers.

An important infrastructural consideration arises from the fact that assessing the clinical significance of genomic information requires widespread access to annotated data about DNA sequence variants and precise, standardized, clinical information about the individuals who have been tested. In recognition of this, the National Center for Biotechnology Information (NCBI) is currently building a freely accessible, public

archive of reports of the relationships between human phenotypes and genotypes, called ClinVar (http://www.ncbi.nlm.nih.gov/clinvar/). At this time, any commercial genetics laboratory depositing data into the resource is doing so on a voluntary basis. Going forward, it may be possible to use the ACA framework of incentives and value-based reimbursement to compel all laboratories to publicly deposit their (anonymized) genetic data into ClinVar or other open access sources by making CMS reimbursement contingent on such behavior, as has been suggested in Europe [45].

ii. Addressing the Health Care Workforce

The ACA recognizes that the changes envisioned through its implementation will require an expanded workforce. Particular emphasis is placed on addressing the increased reliance on primary care but also recognizes the need to address public health, interdisciplinary teams, increased diversity of the workforce and the nursing shortage.

There is also a need to address the disciplines of genetics and genomics, which are rapidly expanding beyond the realm of rare, inherited diseases. However, most health professionals still lack a robust understanding of how to apply and interpret genetic results in clinical situations. Numerous studies document that a majority of health care professionals rate their own genetics knowledge to be "fair" or "poor" and perform inadequately on standardized assessments of genetics knowledge [46]. Among primary care physicians in particular, the lack of genetics knowledge can lead to inadequate or inappropriate referrals, contributing to poor care coordination and unnecessary health care costs. In one recent survey of 5,915 individuals with genetic conditions, 64% claimed they had not received any genetics education materials from the provider most centrally involved in managing their health condition [47]. Training the broader workforce of health care professionals in genetics is crucial to avoid the adverse outcomes of inappropriate genetic testing, including inappropriate referrals and unnecessary health care spending [48].

E. Service Delivery Reform: A Summary

Many heritable conditions involve multi-systemic health concerns that occur throughout an affected individual's life. If the ACA's attempts to improve care coordination, health information technology, and systems integration are successful, they could result in better quality of care for at least some of these individuals, involving not only health care organizations but also school systems and other community-based entities.

The ACA's focus on quality improvement and patient-centered outcomes is an opportunity to develop an evidence base and outcome measures relevant to the health and quality of life goals most important to families with heritable disorders. Patient safety guidelines relevant to populations affected by specific conditions should be formulated and disseminated to local PSOs and other entities concerned with enhancing patient safety and minimizing avoidable adverse events during acute care visits.

The ACA's focus on PCMHs is an opportunity to improve overall care for patients with heritable diseases, given the prior demonstrated success of this model in caring for children and youth with special health needs. To capitalize upon existing PCMH efforts, heritable disease advocates should consider how existing condition-specific organizations and support groups can partner with PCMHs to achieve synergies from existing resources.

Several characteristics of genetic and genomic services will pose financial and logistical challenges during implementation stages of delivery system reforms, especially given the rapid growth of NGS testing and the accelerated pace of scientific advancement in genetics and genomics.

Given the ACA's extensive use of pay-for-performance incentives to motivate service delivery reform, it may be beneficial for advocates of families with heritable diseases to develop and validate standardized quality assessment tools that use performance measures relevant to members of this community. Sources of disparity in

health care access and utilization may differ in this population from those observed in other disease populations and should be monitored prospectively if possible.

The supply and distribution of dedicated medical genetics providers and the genetics knowledge of other health care providers are limited. Efforts to educate the health care workforce in genetics and genomics should target settings and professionals who are likely to have the largest impact on patient referrals and overall care coordination, such as primary care physicians and nurse practitioners.

F. Service Delivery Reform Efforts in Maryland to Date

Although overall physician capacity in Maryland is ample, the state's health care market is dominated by large hospital systems and significant geographic disparity in the distribution of primary care physicians. While Baltimore city is able to boast two major academic centers with excellent pediatric facilities, there are counties in Southern Maryland and on the Eastern Shore, which do not have primary care pediatricians or pediatric inpatient beds. The relative paucity of primary care providers in Maryland is concerning given that approximately 360,000 individuals are expected to be newly insured in Maryland by the year 2020. To ameliorate this situation, Maryland's service delivery reforms have focused on policies enabling health care workforce expansion and encouraging the growth of PCMHs.

i. Patient-Centered Medical Homes in Maryland

Maryland's PCMH program has paid more than $3 million in support payments to primary care practices throughout the state in exchange for their commitment to apply for PCMH recognition from NCQA. The state also began operating a 3-year pilot program to test the PCMH model in 2011, involving 52 health centers across the state, 10 of which are pediatric practices. All major insurance carriers in Maryland are providing enhanced payments to these pilot practices, which are being further rewarded with bonus payments for strong performance according to NCQA metrics.

It remains to be seen whether these efforts will benefit families affected by heritable conditions. This is reflected in data showing that In a 2009-2010 survey, only 62% of parents with special needs children felt that their child's doctors communicated well, indicating significant room for improvement in this regard [54]. A specific concern is that many heritable disease specialists remain affiliated with large academic medical centers, funded in part by NIH research grants. This affiliation raises concerns with whether adequate coordination will occur with various PCMHs that do not have a relationship with those academic medical centers. There is also a concern that since adult onset chronic diseases are so much more common and costly, CSHCN and more specifically those with heritable conditions get lost in the shuffle.

In Maryland, there is an advocacy effort to analyze data pertaining to CSHCN to determine if there would be a large enough group and potential savings. In addition, Maryland has developed its own behavioral health integration effort linking substance abuse, mental health and primary care. A workgroup spent 2012 developing recommendations and implementation will begin over the next several months to a year.

Finally Maryland is addressing development of a new payment and service delivery system as one of 16 states that received a State Innovation Model grant This will entail development of a Health Care Innovation plan to design a new payment and service delivery model in the context of larger health system transformation. The stakeholder meetings for this project have just begun, and the focus has been on adult chronic diseases. The Maryland chapter of the American Academy of Pediatrics is one of the invited stakeholders, so this project may provide an opportunity for input regarding children and adults with congenital disorders.

ii. Health Care Workforce Expansion in Maryland

The Governor's Workforce Investment Board (GWIB) is a steering committee funded by a $150,000 HHS grant to devise a strategic plan for augmenting Maryland's health care workforce in response to the increased demand for primary care services expected to result from health care coverage expansions under the ACA. The GWIB plans to use a mixture of short-term and long-term strategies for investing in Maryland's

health care workforce development, including licensure process improvements, student loan repayment reforms, and changes in workforce liability policies. Although the GWIB has solicited input from a large number of professional groups related to the practice and training of primary health care providers, they do not appear to be focusing explicitly on training needs relevant to medical genetics or heritable disorders [49].

SECTION 4: CONCLUSIONS

The ACA is a complex piece of legislation with broad implications for insurance coverage, benefits and how care will be provided in a "reformed" system. Its impacts will be manifest in major changes in the organization and delivery of services. For persons with heritable disorders and those who work to ensure they receive the services they need, analysis and ongoing surveillance of the implementation of the components of the law is critical. While some issues are relevant to broader populations, there are others specific and/or more critical to this population. Throughout this paper, a number of challenges have been identified that require specific actions and efforts to address the unique needs of persons with heritable diseases.

The economics and logistics of providing genetic services is a longstanding challenge. It is recognized that several characteristics of clinical genetic services limit their potential to achieve economic efficiency [50]. Clinical genetics and genomics are two relatively new medical specialties. They rely on molecular tools that do not lend themselves to evaluation according to traditional technology assessment criteria. Many clinical genetic services are preventive or designed to pre-empt symptoms or truncate lengthy diagnostic odysseys. It is difficult to measure the financial impact of events that were averted as a result of genetic risk assessment. Also, the increasingly rapid pace of scientific advancement in the fields of genetics and genomics has rendered it difficult for payers and providers to keep informed about the latest evidence. These features of clinical genetics function as barriers to securing regular patterns of reimbursement for genetic services.

Medical geneticists vary in their practice of managing patient care on a long-term basis. Overall, it is challenging to define the attributes of an "average" genetics clinic,

given that some medical geneticists are generalists while others devote their entire careers to a single disorder. This dichotomy further complicates matters for payers trying to define adequate coverage policies for genetic services.

Importantly, many components of genetic services involve neither medical procedures nor laboratory testing. Genetic counseling is a time-consuming endeavor, involving labor-intensive case management before and after a patient visit, particularly given the administrative steps often required to secure reimbursement for genetic testing and counseling in the first place [51] [52]. Furthermore, in genetics clinic "the patient" is rarely one person. Genetic services often involve whole families, but the costs of examining and counseling family members of affected individuals are seldom billed.

The growing use of next-generation sequencing (NGS) panels that are not unique to persons with heritable conditions, nonetheless are important issues. NGS can reveal several disease-associated gene variants in a single patient, increasing the time spent on researching and interpreting test results [53]. As NGS testing becomes cheaper and more widely available, cost-effectiveness frameworks will need to consider the prevalence and penetrance of variants identified, the costs of follow-up referrals necessitated by discoveries of incidental genetic test findings, and the potential benefits secured by pursuing genomic testing instead of more cumbersome and invasive diagnostic tests [54] [55].

There are a number of issues that need to be addressed when considering financial and delivery system reform. While many of these issues are generic, the following areas need to be considered while taking into account the specific issues of persons with heritable disorders as well as the increasing individualization of services and interventions needed.

- Identification of the circumstances and areas where benchmark plans are expected to fall short, so that policies for supplemental coverage can be proposed. The case for specific “essential health benefits” and essential community providers must be made.

- The role of state mandates in relation to ACA implementation is unclear at this time, as is the fact that these mandates are not mandatory for self-funded plans. For example, in Maryland over 65% of CSHCN have coverage through self-funded plans. A regional inventory of state mandates that benefit families with heritable conditions is needed to support efforts to ensure they are upheld or incorporated into state-level ACA implementation plans.

- The uncertainties with regard to cost sharing for families leave open the possibility that those with insurance will still face significant financial barriers to care.

- The unique relationship between primary care and specialists for persons with heritable diseases demonstrates the need for increased coordination of their roles. These need to be recognized in the context of new payment methods and delivery approaches. In particular, access to specialty services may require crossing state borders. The structure of current Medicaid programs is a major barrier since services outside the home state may not be covered.

These are complex issues, some of which are being examined in the context of a series of demonstration projects funded under the ACA and managed by the CMS Innovation Center. The demonstrations reflect the need for a more extensive body of knowledge and evidence-based practice to support the goals of improved care and reduced costs. The variety of projects also reflects the potential points of intervention. Currently the primary emphasis is on high-cost users of Medicare and Medicaid. While a recent announcement from the Innovation Center was issued to address financial and service delivery reforms for Medicaid and CHIP populations, there is limited support for efforts that target children in general and, more specifically, persons with heritable diseases.

This new opportunity addressing Medicaid and CHIP could support the case that needs to be made for these underrepresented groups; for the specific interventions for which there is an evidence base and efforts to build a stronger evidence base. However, these is also a need to examine the current demonstrations to identify potential opportunities for learning that will support building that evidence base for persons with heritable diseases.

For example, the current approaches to payment for "care - episodes" needs to address such issues as the integration of genetics into primary care. The services provided by nurses, genetic counselors, nutritionists and other allied health care providers during these care-episodes must be included in the basis for payment. The complex health care needs of families with heritable diseases, as well as appropriate outcome measures should be defined. That will allow adequate payment formulas to be defined, whether as incentives or payment bundles. Building on current CMS projects described earlier as well as potential research efforts through PCORI can help build and further expand the existing evidence-base and address the dynamic nature of the increased knowledge due to new genomics discoveries.

What are the implications for NYMAC and its stakeholders? First and foremost there is a need to be informed about the broad aspects of ACA implementation and more specifically, state-specific efforts since many of the actions are at the state level. Second, identification of opportunities to participate in various implementation activities should be identified and pursued. This includes committees that may be set up to develop and/or advise the states on issues. It might also mean identifying opportunities to testify or provide information to decision-makers.

While major concerns are the content and specifics of the essential health benefit packages, there are many other areas that need to be considered. For example, a review of the training of patient navigators and other assistance to ensure information specific to the needs of families with heritable diseases is included. Providers need to be kept aware of the development of more coordinated approaches to care and payment. . Consumer education about changes is a priority so they are able to receive the care they need.

Recent polls indicate that there is major confusion among the general public. What are the implications for families with special needs and how might they be addressed? They need to be informed to learn both what is happening in ACA implementation and how to mount efforts to support their interests.

At the national level, the National Coordinating Council and the regional collaboratives including NYMAC are being asked to identify actions they can take to help assure that the needs of its target populations will be addressed. The development of this paper and its companion matrix are among the first activities NYMAC has undertaken. A review of current projects has also helped identify areas where there are current activities that may support implementation, especially issues related to service delivery.

As a next step to address the development of a broad strategy and set of activities for the NYMAC region, we propose the establishment of a workgroup. It is anticipated that their efforts will include identification and development of information and materials that can support targeted activities by our stakeholders. Examples may include briefing documents and talking points that can be used by NYMAC stakeholders. It is anticipated that the group will include representatives from each state.

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