

Pursuant to the authority vested in the Public Health and Health Planning Council by sections 206(1)(d), 225(5)(t), and 2733 of the Public Health Law, sections 22.3 and 22.9 of Title 10 (Health) of the Official Compilation of Codes, Rules and Regulations of the State of New York are amended, to be effective upon publication of a Notice of Adoption in the New York State Register, to read as follows:

§ 22.3 - Supplementary reports of certain birth defects [congenital anomalies] for epidemiological surveillance; filing.

(a) Every physician, nurse practitioner authorized to diagnose birth defects, physician assistant authorized to diagnose birth defects, midwife, and hospital as defined in Article 28 of the Public Health Law, [in attendance on an individual diagnosed within two years of birth] providing health care to a pregnant woman or a child under two years of age, who diagnoses an embryo, fetus or child as having one or more of the birth defects [congenital anomalies] listed in Table 1 of this section shall file a supplementary report with the State Commissioner of Health within 10 days of diagnosis thereof.

(b) Every physician, nurse practitioner authorized to diagnose birth defects, physician assistant authorized to diagnose birth defects, midwife, and hospital as defined in Article 28 of the Public Health Law, providing health care to a pregnant woman or a child under ten years of age, who diagnoses an embryo, fetus or child as having one or more of the birth defects listed in Table 2 of this section shall file a supplementary report with the State Commissioner of Health within 10 days of diagnosis thereof.

(c) Every clinical laboratory that conducts diagnostic testing on New York State residents to detect or confirm the diagnosis of genetic or chromosomal anomalies listed in Tables 1 and 2 shall, upon detecting or confirming such a genetic anomaly, file a supplementary report with the State Commissioner of Health within 30 days of detection or confirmation.

(d) Such report shall be on such forms, which may include electronic forms, as may be prescribed by the commissioner to facilitate epidemiological investigation and surveillance.

[Anencephalus and similar anomalies

Spina bifida

Congenital anomalies of the nervous system

Congenital anomalies of the eye

Congenital anomalies of ear, face, neck

Congenital anomalies of heart

Congenital anomalies of circulatory system

Congenital anomalies of respiratory system

Cleft palate and cleft lip

Congenital anomalies of upper alimentary tract

Congenital anomalies of digestive system

Congenital anomalies of urinary system

Congenital anomalies of genital organs

Congenital anomalies of limbs  
 Congenital musculoskeletal deformities  
 Other congenital musculoskeletal anomalies  
 Congenital anomalies of the integument  
 Congenital anomalies of the spleen  
 Congenital anomalies of the adrenal gland  
 Congenital anomalies of other endocrine glands  
 Multiple congenital anomalies  
 anomaly, multiple NOS  
 deformity, multiple NOS]

<b><u>TABLE 1 – BIRTH DEFECTS AND GENETIC DISEASES FOR WHICH REPORTING IS REQUIRED TO AGE 2</u></b>
<u>Malignant neoplasm of kidney</u>
<u>Malignant neoplasm of eye</u>
<u>Malignant neoplasm of brain</u>
<u>Malignant neoplasm of other endocrine systems</u>
<u>Congenital leukemia</u>
<u>Hemangioma</u>
<u>Lymphangioma</u>
<u>Neurofibromatosis</u>
<u>Teratoma</u>
<u>Congenital hypothyroidism</u>
<u>Disorders of thyroid, congenital and hereditary</u>
<u>Diabetes Mellitus, neonatal</u>
<u>Disorders of the pituitary gland, congenital and hereditary</u>
<u>Adrenogenital syndrome</u>
<u>Testicular dysfunction, congenital and hereditary</u>
<u>Dwarfism</u>
<u>Other congenital endocrine disorders</u>
<u>Metabolic and Immunity Disorders, congenital and hereditary</u>
<u>Hereditary Hemolytic anemias</u>
<u>Aplastic anemias, congenital and hereditary</u>

<u>Coagulation defects, congenital and hereditary</u>
<u>Primary thrombocytopenia, congenital and hereditary</u>
<u>Diseases of white cells, congenital and hereditary</u>
<u>Methemoglobinemia, congenital and hereditary</u>
<u>Hereditary diseases of the central nervous system</u>
<u>Extrapyramidal disease and abnormal movement disorders, congenital and hereditary</u>
<u>Spinocerebellar Disease, congenital and hereditary</u>
<u>Anterior horn cell disease, congenital and hereditary</u>
<u>Infantile cerebral palsy</u>
<u>Infantile spasms</u>
<u>Cerebral cysts, congenital</u>
<u>Multiple cranial nerve palsies, congenital</u>
<u>Hereditary peripheral neuropathy</u>
<u>Hereditary muscular dystrophies and other myopathies</u>
<u>Hereditary optic atrophy</u>
<u>Duane's syndrome</u>
<u>Endocardial fibroelastosis</u>
<u>Wolf-Parkinson-White syndrome</u>
<u>Major anomalies of jaw size</u>
<u>Inguinal hernia</u>
<u>Femoral hernia</u>
<u>Nephrotic syndrome, congenital</u>
<u>Nephrogenic diabetes insipidus, congenital</u>
<u>Dyschromia, congenital</u>
<u>Anencephalus and similar anomalies</u>
<u>Spina bifida</u>
<u>Birth defects of the nervous system</u>
<u>Birth defects of the eye</u>
<u>Birth defects of the ear, face, neck</u>
<u>Birth defects of the heart</u>
<u>Birth defects of the circulatory system</u>
<u>Birth defects of the respiratory system</u>
<u>Cleft palate and cleft lip</u>
<u>Birth defects of the upper alimentary tract</u>
<u>Birth defects of the digestive system</u>
<u>Birth defects of the urinary system</u>
<u>Birth defects of the genital organs</u>
<u>Birth defects of the limbs</u>
<u>Congenital musculoskeletal deformities</u>
<u>Other congenital musculoskeletal anomalies</u>
<u>Birth defects of the integument</u>
<u>Birth defects of the spleen</u>
<u>Birth defects of the adrenal gland</u>
<u>Birth defects of other endocrine glands</u>

<u>Multiple birth defects</u>
<u>Anomaly, multiple, Not Otherwise Specified</u>
<u>Deformity, multiple, Not Otherwise Specified</u>
<u>Genetic anomalies</u>
<u>Chromosomal anomalies</u>
<u>Fetal Alcohol Syndrome</u>
<u>Situs Inversus</u>
<u>Conjoined twins</u>
<u>Hamartoses</u>
<u>Birth defect syndromes affecting multiple systems</u>
<u>Noxious influences affecting the fetus via placenta</u>
<u>Amniotic band syndrome</u>
<u>Infections specific to the perinatal period</u>
<u>Hemolytic disease due to RH isoimmunization</u>
<u>Neonatal hepatitis</u>

<b><u>TABLE 2 – BIRTH DEFECTS AND GENETIC DISEASES FOR WHICH REPORTING IS REQUIRED TO AGE 10</u></b>
<u>Hereditary muscular dystrophies and other myopathies</u>
<u>Birth defects of the heart</u>
<u>Genetic anomalies</u>
<u>Chromosomal anomalies</u>
<u>Fetal Alcohol Syndrome</u>

§ 22.9 – Reports: place of filing

All reports required by Section 22.3 of this Part shall be filed with the Director of the Bureau of Environmental [Epidemiology] and Occupational Epidemiology, Center for Environmental Health, [Division of Epidemiology,] New York State Department of Health, Empire State Plaza, Corning Tower [Building], Albany, NY 12237.

## **REGULATORY IMPACT STATEMENT SUMMARY**

### **Statutory Authority:**

Section 206(1)(d) of the Public Health Law (PHL) authorizes the Commissioner to investigate the causes of diseases, epidemics, and the sources of mortality in New York State. PHL § 225(5)(t) provides that the State Sanitary Code may facilitate epidemiological research into the prevention of environmentally related diseases and require reporting of such diseases by physicians, medical facilities and clinical laboratories. PHL § 2733 requires that birth defects and genetic diseases be reported by physicians, hospitals, and persons in attendance at birth in a manner prescribed by the Commissioner. Information collected pursuant to such reports shall be kept confidential pursuant the Personal Privacy Protection Act.

### **Legislative Objectives:**

PHL § 206(1)(d) established the Commissioner's broad authority to investigate the causes of disease in New York State. As reflected in the Declaration of Policy, the Legislature enacted PHL § 2733 and related statutes to ensure that the Department maintains a central and comprehensive responsibility for developing and administering the State's policy with respect to scientific investigations and research concerning the causes, prevention, treatment and cure of birth defects and genetic and allied diseases. Finally, in enacting PHL § 225(5)(t), the Legislature directed that the State Sanitary Code contain regulations that facilitate epidemiological research into the prevention of environmental diseases, by pathological conditions of the body or mind resulting from

contact with toxins, mutagens or teratogens and by requiring the reporting of such diseases or suspected cases of such diseases to the Department.

To these ends, the Department maintains the Congenital Malformation Registry (CMR) and has issued regulations requiring the reporting of structural, functional or biochemical abnormalities determined genetically or induced during gestation, and which are not due to birthing events.

**Needs and Benefits:**

The Department's proposal seeks to extend the case capture periods for certain diseases. Currently, health regulations require physicians and hospitals to report birth defects that are diagnosed within two years of a child's birth, yet many birth defects are not diagnosed until after age two. By extending the capture period for certain diseases listed below, the Department's proposal will enhance its epidemiologic surveillance and advance its understanding of birth defects and their environmental causes.

Fetal alcohol syndrome (FAS) is a serious but preventable birth defect that results from heavy maternal intake of alcohol during pregnancy. FAS is not uncommon, with national estimates of 5–20 cases per 10,000 live births. The annual prevalence of FAS reported by the CMR is about 10-fold less than national estimates. Studies indicate that FAS is more easily diagnosed from ages two to ten years.

Hereditary muscular dystrophies and other myopathies are a family of diseases that cause progressive and steady muscle weakness and wasting. The most common muscular dystrophy is Duchenne MD, followed by Becker MD. A recent US study indicated the prevalence of boys age 5 to 24 with Duchenne and Becker MD was 1.3 to

1.8 per 10,000 males. However, the CMR indicated an annual birth prevalence of only 0.08 per 10,000 live births. One study reported a mean age of diagnosis of 5 years for boys with Duchenne MD.

Congenital heart defects (CHDs) are the most common organ system malformations, and they remain the leading cause of infant deaths from birth defects. Approximately 1 out of every 115 to 150 babies is born with a heart defect. Minor defects are often not detected until later in life and can have serious consequences. One study indicates that 3% of children with CHDs are diagnosed from ages three to ten years old.

Genetic and Chromosomal Anomalies. The CMR was established prior to the sequencing of the human genome and the associated advances in the scientific community's understanding of the role genetics plays in causing birth defects. Because the field of genetics and birth defects is so new, there is little or no documentation about diagnostic timing for many of these syndromes. However, genetic and chromosomal anomalies are often not recognized until after two years of age, because it can require several years to observe a child prior to diagnosis.

The Department's proposal would also require reporting of birth defects diagnosed or identified during pregnancy. This reporting requirement is important due to the increase in routine prenatal screening. For many diseases, the CMR data suggests a prevalence rate in New York that is far below the expected range.

The proposed amendment also allows reporting by qualified health care professionals other than physicians—specifically, nurse practitioners and physician



assistants. Over the past several years, a growing number of national, state and specialty-specific studies indicate that the physician workforce in the United States is facing current and future shortages. Moreover, the shortage of family physicians will be most acute in rural and underserved populations. These trends highlight the need to allow reporting by nurse practitioners and physician assistants. Indeed, anecdotal reports indicate that nurse practitioners and physician assistants are already filling this role because of the burden on physicians.

The regulation would also clarify the requirement that clinical laboratories performing diagnostic testing for birth defects must report to the CMR. This requirement is not new. In 1978, Commissioner Whalen issued a blanket order directing that all laboratories report birth defects to the Department pursuant to PHL § 2733. However, many clinical laboratories are not aware of the reporting requirement.

The Department's proposal adds granularity to the list of reportable diseases. Many diseases currently reported fall under broad categories, thereby limiting the Department's ability to receive information concerning the individual diseases within the category. For example, congenital leukemia and lymphangiomas are both currently reported under the broad classification of "congenital anomalies of the circulatory system." The Department's proposal lists these and other defects as separate reportable conditions.

Finally, the proposal replaces the term "congenital malformation" in favor of the term "birth defect" and renames the CMR the "New York Birth Defects Monitoring

Program.” In a nationally representative survey conducted in 2007, respondents were asked what their first choice would be to describe problems at birth that can result in physical or mental disabilities. The preferred term was “birth defects”. This term was chosen over congenital malformations and congenital anomalies, among other choices. Using the term that is preferred by the public will enable positive engagement with affected families and improve the Department’s communication with the public.

**Costs to Regulated Parties:**

The Department anticipates that, for the entire State, the regulatory changes will require annual reporting of an approximate additional 900 live born children by physicians, nurse practitioners, physician assistants, midwives, and hospitals (FAS: 100-200 cases; muscular dystrophy: 100 cases; cardiac heart defects in children past age two: 200 cases; genetic or chromosomal anomalies: 400 cases).

Approximately 160 New York hospitals and their associated physicians, nurse practitioners, physician assistants and midwives will be affected by this change. The Department anticipates that the costs to these parties will be minimal, primarily because the number of additional birth defects to be reported annually through hospitals (five to six cases per year, on average) will be small, relative to the number of reports already being submitted. Hospitals already report cases to the CMR electronically. The additional hospital staff time to enter six to seven additional cases per year may require 20-30 minutes annually. Alternatively, a hospital can incorporate the additional diagnoses into a monthly batch file. Hospitals are already familiar with the process of modifying batch files.

Reporting by smaller, community-based health care facilities and individual providers will result in some costs primarily because, while physicians have always been required to report birth defects, this requirement has not been enforced for providers who are not associated with New York hospitals. The Department has minimized the administrative costs associated with the reporting requirement by integrating the reporting process with technologies that healthcare providers already utilize. Healthcare providers currently rely on the Department's Health Commerce System (HCS) for communication and reporting to the Department. Within the HCS, the Department is implementing a comprehensive web-based reporting system known as the Child Health Information Integration (CHI<sup>2</sup>) project to be used as the central website to report and track newborn screening, immunizations, lead and newborn hearing screening. Reporting of birth defects will become a component of the CHI<sup>2</sup> system in order to reduce the reporting burden of community-based healthcare facilities and providers.

Providers will be required to spend 3-5 minutes entering case information for each child or fetus diagnosed with a birth defect that is newly reportable under the updated CMR regulations. Statistically, this should involve very few cases for such providers. Because most providers already use and have free access to the online electronic reporting system, the proposed regulation will not impose any additional equipment or technology costs. The only costs will be in the amount of time required to use the CHI<sup>2</sup> to report additional birth defects, which is expected to be negligible. The Department will assist any providers that currently do not have access to the web based reporting system.

With regards to extending the CMR reporting requirements to nurse practitioners, physician assistants and midwives, the Department does not expect that regulated parties

will incur any associated direct costs. Rather, the Department expects that this change will relieve physicians and hospitals from being the only classes of healthcare providers authorized to submit a report when a child is diagnosed with a birth defect.

For clinical laboratories, the Department anticipates the regulatory change will require annual reporting of approximately 6,600 additional genetic or chromosomal anomalies recognized during pregnancy, and approximately 400 reports related to children diagnosed between the ages of 2 and 10 years old, for a total of 7,000 additional reports annually. The Department anticipates the ongoing costs to the roughly 50 clinical cytogenetic laboratories providing diagnostic testing for genetic and chromosomal anomalies to be minimal because these laboratories will report using the Electronic Clinical Laboratory Reporting System (ECLRS) as many already do. The Department estimates that the additional number of reports that these labs will make to ECLRS will cost approximately \$1,400. Clinical laboratories may experience a one-time expense related to modifying the laboratory's software to identify the additional cases that must be reported, which the Department estimates will require a maximum of 16 hours of work by a computer specialist at an estimated rate of pay of \$100/hour.

**Costs to the Regulatory Agency:**

The Department has been using a web-based electronic reporting system in place since 2006. Currently, the CMR receives and processes about 12,000 reports annually. Thus, annual cost to DOH to receive and process the additional 1,000-1,200 cases will be minimal.

**Costs to the State Government:**

There will be no costs to state government. For the last ten years, reporting to the CMR has been conducted electronically. Currently, the Department uses the Health Commerce System to receive CMR reports. Reporters upload cases individually or in batch reports. The electronic reporting system already includes automated processes to match and combine reports for the same child, to ensure de-duplication of data reported from multiple reporters. Additional data quality control processes are built into the system.

**Costs to Local Government:**

Hospitals owned by local governments would be affected but, as discussed above, the costs will be minimal because the additional reporting requirement is relatively small.

**Local Government Mandates:**

There are no mandates on local governments, other than the additional reporting requirements that would apply to hospitals owned by a local government.

**Paperwork:**

This change will generate very little physical paperwork because reporting will be performed electronically as is described under “Costs to Regulated Parties.”

**Duplication:**

This change does not involve any duplication in laws. In terms of duplication of effort, the reporting software will prevent the repeated reporting of the same birth defect for a particular child.

**Alternatives:**

If no changes are made to this regulation, the Department will continue to collect incomplete reporting for birth defects, and prevalence estimates will remain inaccurate. This will impede the Department's ability to detect and quantify environmental exposures that negatively impact the health of embryos and fetuses in New York State.

Concerning FAS, in particular, failure to change the reporting requirement will hamper prevention efforts and may cost New York more in the long-term. One study placed the nationwide annual cost of treating birth defects associated with FAS at \$1.6 billion. Another study used a societal perspective and generated nationwide cost estimates of \$9.69 billion. These costs included estimates of the value of productivity lost as a result of cognitive disabilities, as well as the cost of treatment and residential care. In addition to improving outcomes for affected children, early diagnosis and appropriate interventions are likely to generate significant costs savings over time.

**Federal Standards:**

There are no federal mandates for state-level reporting of birth defects. However, several of the 36 state birth defect surveillance programs require reporting of these birth defects past the age of 2 years, including Hawaii, Texas, Washington State and Colorado. At least eleven states receive reports of birth defects that occur during pregnancy.

**Compliance Schedule:**

Regulations will take effect immediately upon filing. The Department will continue its efforts to make reporting easier and more efficient, while simultaneously conducting outreach to understand and address any concerns that may arise.

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## **REGULATORY IMPACT STATEMENT**

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contact with toxins, mutagens or teratogens and by requiring the reporting of such diseases or suspected cases of such diseases to the Department.

To these ends, the Department maintains the Congenital Malformation Registry (CMR) and has issued regulations requiring the reporting of structural, functional or biochemical abnormalities determined genetically or induced during gestation, and which are not due to birthing events.

**Needs and Benefits:**

The currently proposed amendments to the existing regulation will modernize the CMR in six ways and improve the ability of the CMR to meet the original objectives of the legislation. First, the amendments will establish a case reporting period of ten years for certain defects that often are not diagnosed within the current two-year capture period. The defects subject to the 10-year reporting requirements are: fetal alcohol syndrome (“FAS”); Duchenne and Becker muscular dystrophy; heart malformations; and chromosomal and genetic anomalies.

Second, the amendments will institute case reporting for birth defects diagnosed during pregnancy. Third, the changes will allow reporting from nurse practitioners, physician assistants and midwives, thereby reducing the reporting burden on pediatricians, obstetricians, general and other practitioners and improving reporting in communities where physicians are scarce. Fourth, the amendments will add granularity to the reported data by creating separate categories for certain diseases that are currently reported under a single, broad category. Finally, the changes clarify prior requirements

for clinical cytogenetic laboratories to report diagnostic test results for chromosomal and genetic anomalies.

The Department's proposal seeks to extend the case capture periods for certain diseases. Currently, health regulations require physicians and hospitals to report birth defects that are diagnosed within two years of a child's birth, yet many birth defects are not diagnosed until after age two. In particular, conditions such as fetal alcohol syndrome, Duchenne and Becker muscular dystrophy, certain heart malformations, and some chromosomal and genetic anomalies are not diagnosed until the child is older than two years.

By extending the capture period for certain diseases, the Department's proposal will enhance its epidemiologic surveillance and advance its understanding of birth defects and their environmental causes. The Department's proposal seeks to update the capture periods for the following diseases:

Fetal alcohol syndrome (FAS) is a serious but preventable birth defect that results from heavy maternal intake of alcohol during pregnancy. Significantly, the hallmark signs of FAS are challenging to recognize in infants.

FAS is not uncommon, with national estimates of occurrence at 5–20 cases per 10,000 live births. For birth years 2001 to 2007, the annual prevalence of FAS reported by the CMR was 0.64 per 10,000 live births, about 10-fold less than national estimates. These figures suggest significant underreporting of FAS in New York State. Further, studies indicate that FAS is more easily diagnosed from ages two to ten years. A comparison of the CMR with other FAS surveillance data found that, in one region of

New York State, almost 30% of FAS cases were diagnosed after the age of two. In addition, a recent analysis of children referred to an Erie County FAS Diagnostic Center found the average age of diagnosis of FAS to be 4.9 years, with only 39% diagnosed before their second birthday and 82% diagnosed by a child's tenth birthday. Consistent with current CMR regulations, these diagnoses were not required to be reported to the CMR for children over the age of two. These findings support the Department's proposal to extend the case capture period for FAS to ten years of age.

Hereditary muscular dystrophies and other myopathies are a family of diseases that cause progressive and steady muscle weakness and wasting. The most common muscular dystrophy is Duchenne MD, followed by Becker MD (together, "DBMD"). The age of onset and severity of symptoms are unique for each dystrophy, as is the average age of diagnosis for individuals.

Worldwide, the birth prevalence of Duchenne MD is estimated at 1 to 3.9 per 10,000 live births, and a recent US study indicated the prevalence of boys age 5 to 24 with DBMD for 2007 was 1.3 to 1.8 per 10,000 males. However, the CMR indicated an annual birth prevalence of "hereditary progressive muscular dystrophy" from 1998 to 2007 of only 0.08 per 10,000 live births. The remarkable difference in these statistics suggests significant underreporting of Duchenne MD in New York State.

The statistical difference is likely the result of New York's inadequate two-year case capture period, at least in part. Duchenne MD is commonly not suspected until the child is over two years of age, and one study reported a mean age of diagnosis of 5 years. According to the Department's records, 95% of children were diagnosed with DBMD before their tenth birthday. By increasing the capture period of Duchenne MD to ten

years of age, the Department will improve the accuracy and completeness of its surveillance for this disease, which will help the Department understand the prevalence of this condition and identify regions where healthcare services may be inadequate.

Congenital heart defects (CHDs) are the most common organ system malformations, and they remain the leading cause of infant deaths from birth defects. Approximately 1 out of every 115 to 150 babies is born with a heart defect, compared to only 1 in every 800 to 1,000 babies born with Down syndrome. Variation in prevalence has been associated with maternal race and ethnicity.

Although major heart defects are usually apparent in a newborn, minor defects are often not detected until later in life and can have serious consequences. One study indicates that 70% of children with CHD are diagnosed within the first year of life; an additional 18% are diagnosed in year 2; and 3% are diagnosed up to seven years later. That study found that children with CHD with few or mild symptoms are frequently under-diagnosed, especially in areas with inadequate health services (e.g., lack of nearby tertiary centers and/or cardiology services; insufficient pediatrician awareness and expertise regarding mild malformations). Further, in the past decade, there have been significant advances in medical technologies that can detect mild CHDs in children older than two years of age.

These findings support the Department's proposal to extend the case capture period for CHDs to ten years of age. The improved surveillance will assist the Department's efforts to study causation and support its prevention efforts.

Genetic and Chromosomal Anomalies. The CMR was established prior to the sequencing of the human genome and the associated advances in the scientific

community's understanding of the role genetics plays in causing birth defects. Because the field of genetics and birth defects is so new, there is little or no documentation in the literature about diagnostic timing for many of these syndromes.

However, it is known that genetic and chromosomal anomalies are often not recognized until after two years of age, because it can require several years to observe a child prior to diagnosis. Genetic testing may also be delayed past the age of two because of the cost, insurance policies, or other restrictions related to genetic testing.

By capturing data concerning those children diagnosed with birth defects through genetic and chromosomal testing, the Department will enhance its understanding of the epidemiology of these diseases. The following are two examples of genetic anomalies that will be captured under the CMR's proposed case capture periods.

DiGeorge syndrome ("DGS", also called velocardiofacial syndrome, or VCFS) is a disease that creates cognitive impairments, among other things. DGS has an estimated incidence of 2.5/10,000 live births, yet the CMR's annual birth prevalence of this genetic microdeletion from 1998 to 2007 was only 0.55/10,000 live births. Notably, when a child has minimal facial dysmorphisms, minor cardiac anomalies, and slight cognitive impairments, the child may not be diagnosed within two years of birth. These findings suggest underreporting of DGS in New York State.

The Department believes that the likely underreporting can be remedied, at least in part, by extending the case capture period. A longer capture period, to age 10 years, will dramatically improve the Department's ability to identify children with this disease.

Cystic fibrosis (CF) is the most common life-limiting recessive genetic disorder in Caucasians, with an incidence of 3.1/10,000 live born in the U.S. The reported incidence

has varied from 0.5/10,000 to 3.3/10,000 live births, depending on the population sampled and the method of detection (i.e., newborn screening, newly reported cases, or calculations based on death certificates). However, the CMR's annual birth prevalence of CF from 1998 to 2007 was 1.16 per 10,000 live births. The discrepancy in these statistics likely reflects the CMR's insufficient two-year window of surveillance, which the Department's proposal seeks to address.

The Department's proposal would also require reporting of birth defects diagnosed or identified during pregnancy. This reporting requirement is important due to the increase in routine prenatal screening. For example, observational studies and clinical trials suggest that periconceptual use of folic acid can reduce neural tube defects (NTDs), including anencephaly and spina bifida. In New York State, however, the Department is currently unable to accurately confirm this association or the impact of prevention efforts, because these defects are often diagnosed early in pregnancy and may result in pregnancy outcome other than live birth. Therefore, many NTDs may not be reported to the Congenital Malformations Registry under the current regulations.

In general, for many diseases, the CMR data appears to suggest a prevalence rate in New York that is far below the range of what would be expected, where an approximate expected value is based on data gathered in other states through the National Birth Defects Prevention Network. In particular, CMR data indicates an anencephaly prevalence rate in New York that is approximately 84% less than expected; for anophthalmia, 94% less than expected; Patau syndrome or trisomy 13, 18% less than expected; for Edwards syndrome or trisomy 18, 56% less than expected; spina bifida

without anencephaly, 73% less than expected; and for encephalocele, 44% less than expected. It is highly improbable that the CMR's extremely low prevalence figures reflect the actual prevalence of these diseases in New York State. Rather, the figures are very likely the result of under-reporting by hospitals and healthcare professionals.

These deficiencies in data impede the Department's ability to study the prevalence of birth defects in New York and its relation to environmental factors. The proposed regulatory amendments would correct these deficiencies.

The proposed amendment also allows reporting by qualified health care professionals other than physicians—specifically, nurse practitioners, physician assistants, and midwives. Over the past several years, a growing number of national, state and specialty-specific studies indicate that the physician workforce in the United States is facing current and future shortages. The number of generalist residency graduates and medical students entering primary care has declined each year since 1998. Moreover, the shortage of family physicians will be most acute in rural and underserved populations. These trends highlight the need to allow reporting by nurse practitioners, physician assistants, and midwives. Indeed, anecdotal reports indicate that nurse practitioners, physician assistants, and midwives are already filling this role because of the burden on physicians.

Additionally, reporting by nurse practitioners and physician assistants is key to diagnosing children with fetal alcohol syndrome. Children with fetal alcohol syndrome are more likely to be in foster care settings and covered by Medicaid. Programs serving these children are more likely to employ nurse practitioners and physician assistants

rather than physicians. Thus, to obtain meaningful data concerning fetal alcohol syndrome as well as other birth defects, it is imperative that nurse practitioners and physician assistants be required to report to the CMR.

The regulation would also clarify the requirement that clinical laboratories performing diagnostic testing for birth defects must report to the CMR. This requirement is not new. In 1978, Commissioner Whalen issued a blanket order directing that all laboratories report birth defects to the Department pursuant to PHL § 2733. Although that order remains legally effective, 35 years have passed and many clinical laboratories are not aware of the reporting requirement. Placing the requirement in the regulations will help ensure that clinical laboratories are aware that they must report diagnostic test results to the CMR. This will also support the capture of cases where diagnosis is either occurs during pregnancy or is delayed past age two.

The Department's proposal adds granularity to the list of reportable diseases. Many diseases currently reported fall under broad categories, thereby limiting the Department's ability to receive information concerning the individual diseases within the category. For example, congenital leukemia and lymphangiomas are both currently reported under the broad classification of "congenital anomalies of the circulatory system." The Department's proposal lists these and other defects as separate reportable conditions.



Finally, the proposal replaces the term “congenital malformation” in favor of the term “birth defect” and renames the CMR the “New York Birth Defects Monitoring Program.” In a nationally representative survey conducted in 2007, respondents were asked what their first choice would be to describe problems at birth that can result in physical or mental disabilities. The preferred term was “birth defects”. This term was chosen over congenital malformations and congenital anomalies, among other choices. Using the term that is preferred by the public will enable positive engagement with affected families and improve the Department’s communication with the public.

**Costs to Regulated Parties:**

The Department anticipates that, for the entire State, the regulatory changes will require annual reporting of an approximate additional 900 live born children by physicians, nurse practitioners, physician assistants and hospitals (FAS: 100-200 cases; muscular dystrophy: 100 cases; cardiac heart defects in children past age two: 200 cases; genetic or chromosomal anomalies: 400 cases).

Approximately 160 New York hospitals and their associated physicians, nurse practitioners, physician assistants, and midwives will be affected by this change. The Department anticipates that the costs to these parties will be minimal, primarily because the number of additional birth defects to be reported annually through hospitals (five to six cases per year, on average) will be small, relative to the number of reports already being submitted. Hospitals already report cases to the CMR electronically using one of two methods: by individual child and by batch file. To report a child individually, hospital staff log onto the secure CMR website and enter the required data. It takes about

3 to 5 minutes to enter a complete case. Alternatively, hospitals can submit monthly batch files to the CMR. The additional hospital staff time to enter six to seven additional cases per year may require 20-30 minutes annually. Alternatively, a hospital can incorporate the additional diagnoses into a monthly batch file. Hospitals are already familiar with the process of modifying batch files, so this process is not new or unusual.

Reporting by smaller, community-based health care facilities and individual providers will result in some costs primarily because, while physicians have always been required to report birth defects, this requirement has not been enforced for providers who are not associated with New York hospitals. The Department has minimized the administrative costs associated with the reporting requirement by integrating the reporting process with technologies that healthcare providers already utilize. Healthcare providers currently rely on the Department's Health Commerce System (HCS) for communication and reporting to the Department. Within the HCS, the Department is implementing a comprehensive web-based reporting system known as the Child Health Information Integration (CHI<sup>2</sup>) project to be used as the central website to report and track newborn screening, immunizations, lead and newborn hearing screening. Reporting of birth defects will become a component of the CHI<sup>2</sup> system in order to reduce the reporting burden of community-based healthcare facilities and providers.

Providers will be required to spend 3-5 minutes entering case information for each child or fetus diagnosed with a birth defect that is newly reportable under the updated CMR regulations. Statistically, this should involve very few cases for such providers. Because most providers already use and have free access to the online electronic reporting system, the proposed regulation will not impose any additional equipment or

technology costs. The only costs will be in the amount of time required to use the CHI<sup>2</sup> to report additional birth defects, which is expected to be negligible. The Department will assist any providers that currently do not have access to the web based reporting system.

With regards to extending the CMR reporting requirements to nurse practitioners, physician assistants, and midwives, the Department does not expect that regulated parties will incur any associated direct costs. Rather, the Department expects that this change will relieve physicians and hospitals from being the only classes of healthcare providers authorized to submit a report when a child is diagnosed with a birth defect.

For clinical laboratories, the Department anticipates the regulatory change will require annual reporting of approximately 6,600 additional genetic or chromosomal anomalies recognized during pregnancy, and approximately 400 reports related to children diagnosed between the ages of 2 and 10 years old, for a total of 7,000 additional reports annually. The Department anticipates the ongoing costs to the roughly 50 clinical cytogenetic laboratories providing diagnostic testing for genetic and chromosomal anomalies to be minimal because these laboratories will report using the Electronic Clinical Laboratory Reporting System (ECLRS), which in turn provides appropriate reports to the CMR. These laboratories already use the ECLRS system. The Department estimates that the additional number of reports that these labs will make to ECLRS is expected to cost approximately \$1,400.

Clinical laboratories may experience a one-time expense related to modifying the laboratory's software to identify the additional cases that must be reported. However, the Department estimates that it would require a maximum of two days (16 hours) of work by a computer specialist to modify software to identify the additional cases required by

the regulation for reporting to ECLRS. The estimated rate of pay for a computer specialist is up to \$100/hour.

**Costs to the Regulatory Agency:**

The Department has been using a web-based electronic reporting system in place since 2006. Currently, the CMR receives and processes about 12,000 reports annually. Thus, annual cost to DOH to receive and process the additional 1,000-1,200 cases will be minimal.

**Costs to the State Government:**

There will be no costs to state government. For the last ten years, reporting to the CMR has been conducted electronically. Currently, the Department uses the Health Commerce System to receive CMR reports. Reporters upload cases individually or in batch reports. The electronic reporting system already includes automated processes to match and combine reports for the same child, to ensure de-duplication of data reported from multiple reporters. Additional data quality control processes are built into the system.

**Costs to Local Government:**

Hospitals owned by local governments would be affected but, as discussed above, the costs will be minimal because the additional reporting requirement is relatively small.

**Local Government Mandates:**

There are no mandates on local governments, other than the additional reporting requirements that would apply to hospitals owned by a local government.

**Paperwork:**

This change will generate very little physical paperwork because reporting will be performed electronically. In terms of electronic reporting requirements, physicians, nurse practitioners, physician assistants and hospitals will be required to submit a total of approximately 900 additional reports to the CMR annually. Hospitals already report cases to the CMR electronically using one of two methods: by individual child and by batch file. It takes about 3 to 5 minutes to enter an individual case. On average, hospitals will need to make an additional six to seven reports annually. The additional hospital staff time to enter six to seven additional cases per year may require 20-30 minutes annually. Alternatively, a hospital can incorporate the additional diagnoses into a monthly batch file, which will save some time. Hospitals are already familiar with the process of modifying batch files, so this process is not new or unusual.

Smaller healthcare providers will also be required to spend 3-5 minutes entering case information for each child or fetus diagnosed with a birth defect that is newly reportable under the updated CMR regulations. Statistically, this should involve very few cases for such providers. The Department will assist any providers that currently do not have access to the web based reporting system.

For all clinical laboratories, the Department anticipates the regulatory change will require annual reporting of approximately an additional 7,000 genetic or chromosomal anomalies. Laboratories will use the Electronic Clinical Laboratory Reporting System

(ECLRS), which in turn provides appropriate reports to the CMR. These laboratories already use the ECLRS system, and the additional number of reports that individual laboratories will make to ECLRS is relatively small and does not represent a significant reporting burden.

**Duplication:**

This change does not involve any duplication in laws. In terms of duplication of effort, the reporting software will prevent the repeated reporting of the same birth defect for a particular child.

**Alternatives:**

If no changes are made to this regulation, the Department will continue to collect incomplete reporting for birth defects, and prevalence estimates will remain inaccurate. This will impede the Department's ability to detect and quantify environmental exposures that negatively impact the health of embryos and fetuses in New York State.

Concerning FAS, in particular, failure to change the reporting requirement will hamper prevention efforts and may cost New York more in the long-term. One study placed the nationwide annual cost of treating birth defects associated with FAS at \$1.6 billion. Another study used a societal perspective and generated nationwide cost estimates of \$9.69 billion. These costs included estimates of the value of productivity lost as a result of cognitive disabilities, as well as the cost of treatment and residential care. In addition to improving outcomes for affected children, early diagnosis and appropriate interventions are likely to generate significant costs savings over time.

**Federal Standards:**

There are no federal mandates for state-level reporting of birth defects. However, several of the 36 state birth defect surveillance programs require reporting of these birth defects past the age of 2 years. For example, FAS must reported at any age in Hawaii, to age six in Texas, and to age 10 in Washington State. In Colorado, reporting of most defects is up to age 3, but reporting of FAS is required up to age 10. Other states have FAS capture periods ranging from 4 to 18 years. At least eleven states receive reports of birth defects that occur during pregnancy.

**Compliance Schedule:**

Regulations will take effect immediately upon filing. The Department will continue its efforts to make reporting easier and more efficient, while simultaneously conducting outreach to understand and address any concerns that may arise.

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**REGULATORY FLEXIBILITY ANALYSIS FOR SMALL BUSINESSES  
AND LOCAL GOVERNMENTS**

**Effect of Rule:**

This amended rule will have limited impact on small businesses providing health care because many of these businesses are affiliated with a general hospital. These small businesses include community-based healthcare providers (pediatricians, family practitioners and maternal-fetal medicine specialists) and some laboratories with small offices.

The amended rule will have a small impact on those healthcare facilities that are owned by local governments and that also diagnose birth defects and genetic diseases. These healthcare facilities will be required to make additional reports to the CMR based on the updated list of reportable birth defects and genetic diseases. Although the Department does not maintain a listing of local government-owned facilities that would be required to report, the Greater NY Hospital Association estimated that the number is relatively few. Further, the Department reasonably expects the burden on such facilities to be small—only 3-5 minutes per additional case. The number of cases will vary depending on the size of the facility, but the Department estimates that such facilities will report an average of 5-6 newly reportable cases per year, per facility.

**Compliance Requirements:**

Because healthcare providers and facilities are transitioning to electronic record-keeping systems, reporting and record keeping are expected to be simple and require very little time. The Department publishes a CMR guide to assist hospitals with reporting. A

guide will also be developed for other healthcare providers as well as clinical laboratories.

**Professional Services:**

No additional professional services are required under the amended rule.

**Compliance Costs:**

Staff working in small community-based healthcare providers and small clinical laboratories will need to learn how to report with the updated CMR requirements.

**Economic and Technological Feasibility:**

The amended rule is economically and technologically feasible because local governments and small businesses that are affected will continue submitting reports using their free access to the Department's electronic reporting system.

**Minimizing Adverse Impact:**

By offering free access to the electronic reporting system, the Department has minimized the costs and impact on local governments and small businesses operating in New York State.

**Small Business and Local Government Participation:**

The Department has reached out to the healthcare community to gather feedback on the proposed amended rule. Those contacted include: NYS American Academy of Pediatrics, NYS Academy of Family Physicians, Nurse Practitioner Association of NYS, NYS Nurses Association, NYS Society of Physician Assistants, NY Health Information Management Association, Greater NY Hospital Association, Healthcare Association of NYS, NYS March of Dimes, NYS Clinical Geneticists, Genetic Counselors, Midwives, Neurologists, Neuromuscular Specialists, and Pediatric Cardiologists. Additionally, the

Department contacted other NYS agencies and programs which provide services to children affected by these birth defects, specifically fetal alcohol syndrome.

The Department received comments from two organizations that represent health care providers. The President of the New York State Society of Physician Assistants stated, “After soliciting input from our leadership, we wholeheartedly support this suggested regulatory change.” No concern was expressed about costs. Greater New York Hospital Association (GNYHA), representing nearly 150 voluntary, not-for-profit, and public hospitals expressed concern that “raising the maximum reporting age to 10 ... could potentially create an administrative burden for health care providers ... already contending with a wide range of such requirements.” GNYHA strongly recommended that the DOH work closely with providers to develop and implement a reporting system that places the least possible amount of administrative burden on those impacted by this potential regulatory change.

The Department also received positive support for these regulatory changes from non-profit organizations and other State agencies, including the NYS Council on Children and Families, the NYS Office of Alcoholism and Substance Abuse Services, the NY State Education Department’s Office of Special Education, and the Long Island Council on Alcoholism and Drug Dependence. These organizations view the proposed regulatory change as positive steps for meeting the needs of children and families affected by these devastating birth defects.

The Department asked several maternal-fetal medicine practices for input concerning the proposed changes and received replies from three practices (Hudson Valley Perinatal Consulting, Harrison, NY; University GYN/OB, Inc, at Women and

Children's Hospital of Buffalo, Buffalo, NY; and Fetal Testing Unit of Mercy Hospital Buffalo South, Buffalo, NY). As for access to the Department's web based reporting system, one had access, one did not, and the third was uncertain. All three expressed concerns about time required to report and assurances of patient confidentiality.

The Department reached out to the NYS Association of Licensed Midwives, who supported the amendment. In a survey sent to midwives, all respondents supported the regulatory amendment. The most common concern was the time required to comply, which the Department will minimize through its electronic reporting.

Public Health Law § 206(1)(j) ensures that diagnoses reported to the New York Birth Defects Monitoring Program shall be kept confidential and shall be used solely for the purposes of the Department's scientific research. The statute further provides that such records are not admissible as evidence in a court of law. Regarding time to report, the Department expects that some of these practices may not actually have to report separately but that their associated institution or hospital will be able to assume that responsibility, thus reducing the anticipated burden.

The Department is committed to minimizing the administrative burden of these new reporting requirements. By using the CHI<sup>2</sup> system as a reporting tool, the administrative burden will not be significant.

The Department will continue to communicate with stakeholders throughout the regulatory process. Prior to adoption of the rule, all amendments will appear in the New York State Register for public comment.

## **RURAL AREA FLEXIBILITY ANALYSIS**

### **Types and Estimated Numbers of Rural Areas:**

This regulation would apply statewide and affect the 44 counties that are considered rural.

### **Reporting, Recordkeeping and Other Compliance Requirements; and Professional Services:**

This change involves a small increase in reporting using a system already being utilized by healthcare professionals to submit other reports. No additional requirement for professional services is required under the amended regulation.

### **Costs:**

There is minimal cost to report. The costs are associated with staff time to report additional cases electronically. The number of additional cases to be reported is expected to be small relative to the number of cases already reported.

### **Minimizing Adverse Impact:**

Any adverse impact will be minimized by using the Department's pre-existing Health Commerce System for electronic reporting. The impact will be further reduced when the Department implements the CHI<sup>2</sup> reporting system.

### **Rural Area Participation:**

Regulated parties in rural areas have been contacted through the Department's reaching out to statewide associations of healthcare professionals, such as the NYS American Academy of Pediatrics, NYS Academy of Family Physicians, Nurse Practitioner Association of NYS, NYS Nurses Association, NYS Society of Physician

Assistants, NY Health Information Management Association, Healthcare Association of NYS, NYS March of Dimes, and NYS Clinical Geneticists.

## **JOB IMPACT STATEMENT**

### **Nature of Impact:**

There will be minimal impact, because health care facilities are currently required to report other conditions to the Department of Health. The Department does not expect there to be a positive or negative impact on jobs or employment opportunities.

### **Categories and Numbers Affected:**

The Department anticipates no negative impact on jobs or employment opportunities as a result of the amended rule.

### **Regions of Adverse Impact:**

The Department anticipates no negative impact on jobs or employment opportunities in any particular region of the state.

### **Minimizing Adverse Impact:**

Not applicable.