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Newborn Metabolic Screening in Alaska

Newborn metabolic screening was first implemented to find infants born with Phenylketonuria (PKU). Over time, screening panels have expanded to include other disorders. Since 1967, Alaska has attempted to screen all infants born in the State for heritable diseases that can lead to mental retardation or death if left untreated. The Alaska Newborn Metabolic Screening Program tracks infants for screening and surveillance to ensure that infants needing further diagnostic testing are evaluated, treated, and referred to the Alaska Genetics Program.

Seriousness

Healthy People 2010 Targets and National Data

Indicator	Alaska†	Healthy People 2010 Goals*
Ensure that all newborns are screened at birth for conditions mandated by their State-sponsored newborn screening programs	>99%	Developmental
Ensure that follow-up diagnostic testing for screening positives is performed within an appropriate time period	100%	Developmental
Ensure that infants with diagnosed disorders are enrolled in appropriate service interventions within an appropriate time period	100%	Developmental

Table 1

Incidence Rates of Metabolic Disorders/Diseases		
Disorder/Disease	Alaska 2001-04†	Nation^
	per 10,000 births	
Congenital Hypothyroidism (CH)	5.5	3.3
Biotinidase	0.25	0.17
Maple Syrup Urine Disease (MSUD)	0.5	0.06
Phenylketonuria (PKU)	0.5	1.0
Medium Chain Acyl-CoA Dehydrogenase Deficiency (MCAD)	1.5	1.0
Congenital Adrenal Hyperplasia (CAH)	2.5	0.83
CAH among Yupik Eskimos	33.3	----

- In 2004, 10,221 infants were screened with 12 confirmed cases of disease or a disorder and another 167 identified with an abnormal homozygous hemoglobinopathy, carrier trait, or alpha thalassemia.
- Alaska currently obtains metabolic screens on nearly all infants born in the state. Of these, nearly 90% receive a second metabolic screen.
- Maple Syrup Urine Disease (MSUD), an inherited disease, that if untreated, results in increased mental retardation, physical disability, and death – is more than 8 times higher in Alaska than the estimated national incidence. (Table 1)
- Congenital adrenal hyperplasia (CAH), a genetic defect of the adrenal glands that prohibits the production of corticosteroids, that if untreated, can cause heart failure and death within a few days from birth. CAH in Alaska is 3 times the estimated national incidence. (Table 1)

Severity

Metabolic disorders are rare but collectively have an incidence of approximately 1 in every 1,000 to 3,000 births, nationally.[1] Early treatment and intervention has prevented mental retardation or early death for more than 20 infants born in Alaska in the previous two years.

Urgency

- Many of the disorders in the metabolic panel can be detected on a second screen with a normal first screen (termed late onset disorders). Although nearly 90% of Alaskan newborns received a second screening in 2004 – an increase of 9.4% from the previous year (81.95%), all newborns should receive a second screening.
- In Alaska, 6% of all infants with PKU and 10% of those with hypothyroidism are found only on the second screen.[2]

Disparities

- Alaska Native infants have a higher incidence of CAH. In 2001-2004, the average incidence of CAH in Alaska was more than 13 times higher among Yupik Eskimos compared to Alaska overall. (Table 1)

- While no statistics have been compiled for carnitine disorders to date, all but one of the presumptive and confirmed positives for 2004 were among Alaska Native infants.

Economic Loss

Studies have compared the cost of newborn metabolic screening to the high lifetime costs associated with caring for a developmentally/physically disabled child.[3,4] Studies suggest that newborn metabolic screening for PKU, congenital hypothyroidism, and the rarer disorders result in a large cost savings to society in general. Cost effectiveness for some of the newer conditions included in the metabolic panel has not been evaluated.

Interventions & Recommendations

The March of Dimes has advocated for expanded testing through tandem mass spectrometry[5] as have the Association of Public Health Laboratories who prepared a statement for the Secretary of Health and Human Services for the Secretary's Advisory Committee on Heritable Disorders and Genetic Diseases in Newborns and Children.[6]

Intervention Effectiveness

Data from the Alaska Newborn Metabolic Screening Program conclusively shows that early identification and treatment of metabolic disorders is effective (see 'Severity').

Capacity

Propriety

Newborn metabolic screening to prevent mental retardation is a population based program covering all infants born in the State of Alaska.

Economic Feasibility

There are huge economic consequences that would fall upon the State if infants were not identified at birth with metabolic conditions that can easily be treated. The cost of a metabolic screen for an Alaskan infant is $55

Acceptability

There is universal acceptance of newborn screening.[7] While most parents are aware of the "heel stick test" their babies receive, many do not fully understand how many how which conditions are screened. Efforts to get educational materials about metabolic screening to parents in the prenatal period are ongoing.

Resources

Title V MCH Block Grant

Legality

Metabolic screening is mandatory in Alaska covered under "Article 6. Phenylketonuria" in "Sec.18.15.200 Screening infants for Phenylketonuria" and in additional regulations in "Article 10. Screening of Newborn Children for Metabolic Disorders (7AAC 27.510-7AAC27.590).

References

1 Bryant KG, Horns KM, et. al. A Primer on Newborn Screening. Adv Neonatal Care: 4(5):306-317. 2004.
2 Northwest Regional Newborn Screening Program Practitioner's Manual, 6th Edition. 2003.
3 Dhondt JL, Farriaux JP, et. al. Economic Evaluation of Cost-Benefit Ratio of Neonatal Screening Procedures for Phenylketonuria and Hypothyroidism. J Inherit Metab Dis: 14:633-639. 1991.
4 Brosnan CA, Brosnan P, Therrell, BL, et. al. A Comparative Cost Analysis of Newborn Screening for Classic Congenital Adrenal Hyperplasia in Texas. Pub Health Rep: 113:170-178. 1998.
5 Newborn screening Report. March of Dimes. Available at: www.modimes.org
6 Genetics. Maternal Child Health Bureau, DHHS, HRSA. Available at www.mchb.hrsa.gov

Data Sources

† Alaska Newborn Metabolic Screening Program, 2004 Data: State of Alaska, DHSS, DHCS. www.hss.state.ak.us/dhcs/screening/metabolic

* Healthy People 2010. U.S. Department of Health and Human Services. Healthy People 2010. 2nd ed. With understanding and improving health and objectives for improving health. 2 Vols. Washington, DC: U.S. Government Printing Office. 2000.

‡ Alaska Newborn Metabolic Screening Program, 2001-2004 Data: State of Alaska, DHSS, DHCS. www.hss.state.ak.us/dhcs/screening/metabolic

^ National Incidences of Newborn Metabolic Disorders in: Northwest Regional Newborn Screening Program Practitioner's Manual, 6th Edition. 2003.