State of Alaska Epidemiology Bulletin

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Carnitine Palmitoyl Transferase-1A Deficiency Rates in Alaska

BACKGROUND

According to state statute Sec. 18.15.200 and regulations 7AAC 27.510-.590, all newborn babies in Alaska are required to undergo screening for metabolic disorders. In October 2003, screening was expanded to include more than 30 conditions. Carnitine palmitoyl transferase-1A (CPT-1A) deficiency, one of the newly added conditions, is a rare, autosomal recessive disease that results in defective fatty acid metabolism. Patients with untreated CPT-1A deficiency usually present for medical care after the newborn period with seizures or coma associated with life-threatening episodes of fasting hypoketotic hypoglycemia.

CPT-1A deficiency screening tests for Alaska are performed at the Oregon Public Health Laboratory, using tandem mass spectrometry (TMS) analysis of a heel stick blood spot. Positive screening results are reported to the Alaska Division of Public Health (DPH) to facilitate confirmation of diagnosis. The purpose of this *Bulletin* is to inform health-care providers of the incidence of detected CPT-1A deficiency in Alaska and to provide appropriate follow-up recommendations.

METHODS

We examined CPT-1A deficiency incidence rates among infants born in Alaska after expansion of the metabolic screening panel.

RESULTS

From October 1, 2003 through June 30, 2006, 28,340 births occurred in Alaska and 38 infants with CPT-1A deficiency were reported to DPH (rate, 1.3 per 1,000 live births). All identified cases occurred among Alaska Natives (rate, 5.3 per 1,000 live births). Of the 38 infants with CPT-1A deficiency identified, 24 (63%) lived in Western Alaska, 8 (21%) lived in Northern Alaska, and 6 (16%) lived in other regions of Alaska. Twenty-five (66%) of the infants were male.

DISCUSSION

Patients with CPT-1A deficiency may have some functional CPT-1A enzyme, depending on the type of DNA mutation they have.[1] DNA analyses have found that all of the Alaska Native newborns with CPT-1A deficiency to date have had 10-25% of the normal amount of the enzyme.[2] Many Canadian Inuits have also been found to have the same DNA mutation.[3]

A DNA diagnostic test looking for the P479L founder gene mutation, which is performed with a dried blood spot or a buccal swab, can be used to confirm the diagnosis.[2]

Treatment of this disorder requires frequent feeding of affected infants. Other treatment modalities may include a diet low in fat, high in carbohydrates, and supplemented with medium chain triglyceride oils. If treatment is not implemented and repeated episodes of metabolic crisis occur, there is a chance for permanent learning disabilities or mental retardation. After 5 years of age, metabolic crises tend to happen less frequently and are not as severe. With prompt and careful treatment, children with CPT-1A deficiency often live healthy lives with typical growth and development.

RECOMMENDATIONS

1. Refer CPT-1A-deficient patients to the State Metabolic Genetics Clinic for consultation regarding specific dietary guidelines and follow-up evaluation (call 907-269-3430 during working hours).
2. Consider CPT-1A deficiency in pediatric/adolescent patients-particularly Alaska Natives-who present with hypoketotic hypoglycemia (call the Newborn Metabolic Screening Program at 907-269-3499 for information).
3. Test biological siblings to newly identified CPT-1A-deficient patients, using the DNA diagnostic test.
4. Instruct parents of children with CPT-1A deficiency to follow the age-specific feeding guidelines detailed in the Alaska Department of Health and Social Services CPT-1 Deficiency Brochure.[4]
5. Instruct parents to bring their CPT-1A-deficient infants in for medical evaluation if they have not eaten for ≥8 hours.
6. If a CPT-1A-deficient infant/child requires surgery, administer glucose-containing intravenous fluids before surgery and continue until the child is able to eat.
7. For additional information, contact the Section of Women's, Children's & Family Health at 907-269-3400.

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