

TREATMENT JUSTIFICATION FOR PHENYLKETONURIA (PKU)

ICD.9.CM 270.1

Phenylketonuria (PKU) is a rare metabolic disorder usually due to a defect in the enzyme responsible for the metabolism of phenylalanine (PHE), an essential amino acid. The disorder affects one in approximately 15,000 infants born in the United States. If untreated this disorder causes profound mental retardation and other medical problems. Newborn Screening for PKU is the standard of practice in the United States.

The treatment is the restriction of phenylalanine using medical food products “formulas” as defined in section 5(b) of the Orphan Drug Act (21 U.S.C. 360ee(b)(3)). These foods are formulated to be consumed enterally under the supervision of a physician and intended for the specific dietary management of a disease or condition. “Special medical food products” are food products that are prescribed by a physician for the treatment of PKU and used in place of normal food products such as grocery store foods used by the general population. These “formulas and special food products” are medically necessary for the treatment of PKU. The formulas cannot be obtained “over the counter”.

SB 148 is an act to add Section 1374.56 to the Health and Safety Code and to add Section 10123.89 to the Insurance Code, relating to health care coverage. SB148 mandates coverage for the treatment of PKU which includes those formulas and special foods products that are part of a diet prescribed by a licensed physician and managed by a health care professional in consultation with a physician who specializes in the treatment of metabolic disorders. Willful violation of these provisions is a crime. A copy of this bill is attached for review.