**Section 661.320 Congenital Adrenal Hyperplasia (CAH) (Secondary to 21-hydroxylase deficiency)**

a) Interpretation of Results. Although the majority of infants affected by CAH will be identified by this screening, due to genetic variabilities and variations in health status, specimen quality, and timing of specimen collection, not all infants affected by the disorder may be identified. As with any laboratory test, false positive and false negative results are possible. Newborn screening test results are insufficient information on which to base diagnosis or treatment.

1) Neonatal levels for 17-hydroxyprogesterone vary with gestational age, birth weight, time of collection and in response to concurrent medical problems. Normal 17-hydroxyprogesterone levels shall be established using accepted statistical techniques (for example, as described by the Association of Public Health Laboratories, see Section 660.20).

2) When the 17-hydroxyprogesterone level is deemed to be abnormal, the Department will recommend a repeat newborn blood spot screening test or referral of the newborn to a designated pediatric endocrinologist for further evaluation for CAH.

b) Designation of Medical Specialist. In addition to the minimum qualifications set out in Section 661.230, medical specialists designated by the Department to follow-up on a screen positive for CAH shall possess training in pediatric endocrinology with certification of special competence in pediatric endocrinology by the American Board of Pediatrics.

c) Diagnosis and Treatment. Medical management by a designated pediatric endocrinologist is highly recommended. Replacement therapy with glucocorticoids and, in some cases, mineralocorticoids is currently the standard treatment. Long-term follow-up of children with CAH is necessary to adjust medications and to assess growth and development.