**Section 661.300 Adrenoleukodystrophy (ALD)**

a) Interpretation of Results. Although the majority of infants affected by ALD 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) Using tandem mass spectrometry or other methods, ALD is indicated when an elevation of lysophosphatidylcholine (C26LPC) is detected in dried blood spots.

2) When the C26LPC levels are found to be abnormal, the Department will recommend a repeat newborn screening test or referral to a designated medical specialist for further diagnostic studies.

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 ALD shall be certified by the American Board of Medical Genetics and Genomics in Clinical Biochemical Genetics, Medical Biochemical Genetics, or possess certification by the American Board of Medical Genetics and Genomics in Clinical Genetics with at least one year of post-training experience in the diagnosis and treatment of ALD or other peroxisomal disorders. ALD medical specialists shall have the capacity to provide a multidisciplinary approach to care, including the availability of a pediatric endocrinologist with certification of special competence in Pediatric Endocrinology and a pediatric neurologist with certification of special competence in Pediatric Neurology by the American Board of Pediatrics.

c) Diagnosis and Treatment. Medical management by a designated medical specialist is highly recommended to confirm the diagnosis of ALD and other peroxisomal disorders. Referral to the appropriate medical team, including pediatric endocrinology and pediatric neurology, is critical for monitoring and treatment. Long-term follow-up is necessary to document and to assess growth and development.