

Adrenoleukodystrophy – Follow-up of Positives

	Adrenoleukodystrophy (ALD) Protocol – NBS <i>Metabolic</i>	
	Go-Live	<i>September, 21, 2016 Implementation Date</i>
NAPS Lab	NBS Screening Panel Completed	<ul style="list-style-type: none"> • Transfer specimen to GDL with tracking • Specimen tested for ALD at GDL Lab
GDL ALD Lab	MS/MS for C26:0 Tier 1	<ul style="list-style-type: none"> • Initial VLCFA Tier 1 - Inconclusive NBS Results held pending further testing • Initial VLCFA Tier 1 - Negative NBS Results released to SIS
GDL ALD Lab	HPLC MS/MS for C26:0 Tier 2	<ul style="list-style-type: none"> • VLCFA Tier 2 - Positive NBS Results held pending further testing Ship specimen to Greenwood Genetic Center Lab with tracking • VLCFA Tier 2 - Negative NBS Results released to SIS
Greenwood Genetics Lab	ABCD1 Gene Sequencing Tier 3	<ul style="list-style-type: none"> • Greenwood Genetics Center Lab ABCD1 Gene Sequencing NBS Results held pending sequencing • Sends sequencing result to GDL Lab
GDL ALD Lab	Results released in SIS	<ul style="list-style-type: none"> • NBS Results, Tier 2, and ABCD1 Sequencing Mailers are released when Tier 3 Sequencing complete
ASC	ALD ABCD1 Mutation Positive or ALD ABCD1 Mutation Negative	<ul style="list-style-type: none"> • SIS Headline Case Screen notification to ASC • Contact NICU or PCP within 2 Business Days • Notify of result and need for referral to Metabolic Center. • Fax/Email all result mailers, ALD Fact Sheet and Request Family Notification • Refer to Metabolic Center with TE 161

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		<ul style="list-style-type: none"> • Track 14 days to confirm family connected to Metabolic Center • If the center has difficulty making contact with family, offers assistance in making a referral to public health nursing • Documents all attempts at notification, interactions with physicians and parents using tracking events and case notes in SIS. • Reports unusual occurrences such as missed cases, anomalous/inconsistent results, lost to follow-up cases, delays in contacting family, etc., to the NBS Nurse Consultant/ASC Contract Liaison.
Metabolic Center	ABCD1 - Mutation Positive	<ul style="list-style-type: none"> • Contact family for Initial Visit and Counseling • Initiate additional Confirmatory and baseline testing • Enter initial ALD Service Report in SIS • Recall family per established protocol • Updates Case Resolution Report in SIS
Metabolic Center	ABCD1 - Mutation Negative	<ul style="list-style-type: none"> • Contact family for Initial Visit and Counseling • Initiate additional Diagnostic testing • Enter Case Resolution Report in SIS • Recall family per established protocol • Complete Initial Service Report • Update Case Resolution Report in SIS
ASC	Case Resolution and Closure	<ul style="list-style-type: none"> • Close case per Case Resolution diagnosis