

ALD Screening Data

As of August 21, 2017, 721,492 California babies screened for ALD

	Count	Positive Rate	Positive frequency
Total Tier 1 Positives	19,769	2.7%	1 in 36
Total Tier 2 Positives	158	0.02%	1 in 4,566

Incidence Rate Estimates by Sex and ABCD1 Sequencing Interpretations:

	Known Disease Causing + Variants of Unknown Significance	Known Disease Causing Only
Affected Males/ Total Male Births	1 in 7,800	1 in 32,800
Female Carriers/ Total Female Births	1 in 6,300	1 in 19,000
Total Births	1 in 7,000	1 in 24,100

Findings from DNA Sequencing of 158 Newborns With Elevated Tier 2 ALD Screening Results,
2/2016 –8/2017

Variant Interpretation	Count	Percent
Known Pathogenic		19.0%
Boys	11	
Girls	19	
VOUS		46.2%
Boys	35	
Girls	38	
Benign Polymorphism		21.5%
Boys	10	
Girls	24	
No Mutations		13.3%
Boys	10	
Girls	11	