CTX Awareness Day
September 17, 2025

Dear Friend(s),

On behalf of the CTX Alliance and the hundreds of other families whose lives are affected by the rare disease Cerebrotendinous Xanthomatosis (CTX) I am writing to request that a proclamation be issued this year for CTX Awareness Day, which is on September 17, 2025.

CTX is a rare, progressive, genetic lipid storage disorder that is autosomal recessive in nature. Patients affected with CTX have a deficiency in the enzyme sterol 27-hydroxylase, which is encoded by the CYP27A1 gene. Disease-causing variants in the CYP27A1 gene affect the production of sterol 27-hydroxylase. This enzyme is required to convert cholesterol into bile acids, which are important in the absorption of fats and fat-soluble vitamins in the intestine. Sterol 27-hydroxylase deficiency results in impaired synthesis of chenodeoxycholic acid (CDCA), a primary bile acid, leading to a disrupted bile acid metabolism and toxic overaccumulation leads to cholestasis, cataracts, diarrhea, xanthomas, learning difficulties, neuropsychiatric issues, as well as ataxia, cognitive impairment, and progressive neurologic deterioration.

Only 5% of rare diseases have an FDA approved treatment or cure and CTX is one of them. One of the biggest challenges continues to be diagnosing CTX, and the variability of onset and severity of CTX leads to the underdiagnosis and delayed diagnosis of patients affected. The average age of diagnosis is 35.5 years ±11.8 years, and the average diagnostic delay is 16 years. As a rare disease, raising awareness is one of our greatest tools to help individuals living with this disease. With your support in declaring September 17th CTX Awareness Day, we can continue to increase awareness about this neurodegenerative disease. Your office can make significant changes and use its incredible power and respect to help educate residents about this rare disease. With your support and acknowledgement, you can help patients in our community learn about the disease and end the diagnostic journey for patients who have desperately sought a diagnosis, by accessing testing, and receiving treatment.

Please find below a draft of a proposed proclamation for CTX Awareness Day on September 17th.

Thank you for your time and consideration on behalf of all families affected by this devastating disorder. Together, we can help these patients!

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WHEREAS it is the custom of this Legislative Body to recognize official days to increase awareness of serious illnesses that affect the lives of citizens of Name of State.

WHEREAS a diagnosis of a genetic variant on the CYP27A1 gene means an individual could have neurodevelopmental differences, suffer from chronic diarrhea, bilateral cataracts, seizures, tendon xanthomas, difficulty with coordination and balance, and can experience progressive neurological problems over time.

WHEREAS CYP27A1 is a gene the provides instructions for producing an enzyme that helps to break down cholesterol to form acids used to digest fats. When there is a variant on this gene, the enzyme does not work properly and prevents these fats from breaking down properly, affecting the eyes, tendons, brain, and other tissues over time.

WHEREAS individuals with a variant on the CYP27A1 gene can experience life-altering neurological changes including difficulty with coordination and balance, seizures, and cognitive impairment.

WHEREAS treatment options for individuals with a CYP27A1 gene variant are limited to lifetime use of chenodeoxycholic acid replacement therapy which may partially reverse the disease process but cannot reverse damages that have been caused by it.

WHEREAS it is imperative that there be greater public awareness of this health issue, and more must be done to increase activity at the local, state, and national levels.

NOW, THEREFORE, as Governor of the State of Name of State I do hereby proclaim September 17, 2025, CTX Awareness Day in the Name of State.

Signature: _____ **Date:** _____