Sponsored by the ACD and Johnston family on behalf of Kelly Johnston III

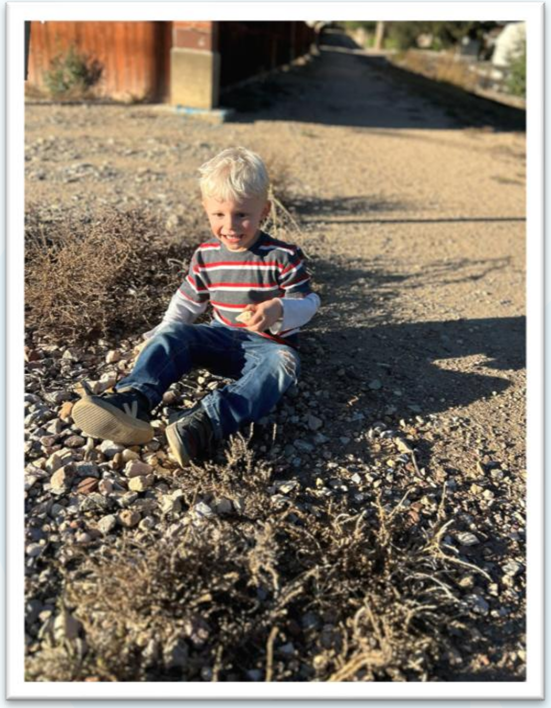
ACD
ASSOCIATION FOR CREATINE DEFICIENCIES

Race for a Cure

by Association for Creatine Deficiencies

Race for the Cure Fundraising Campaign

Kelly III was diagnosed with Creatine Transporter Deficiency (CTD)



- Creatine is an **essential** energy source for normal development and function of the brain, heart and muscles. Our bodies make creatine and we also ingest it through our diet.
- CTD is an **ultra-rare genetic disorder** with <150 documented cases globally. The body does not make the necessary **transporter** to get creatine into cells due to a **single-gene mutation**, SLC6A8. High-energy organs such as the brain cannot function normally without this vital energy source.
- CTD is largely underdiagnosed due to range of symptoms and severity.
- All children with CTD have **severe speech and intellectual delays.**
- CTD is one of 3 different creatine deficiency disorders. Those with GAMT and AGAT are not able make creatine in their bodies, however, do well with supplementation. Kelly does not have the transporter to bring creatine into cells, so **supplementation is unlikely to help**.

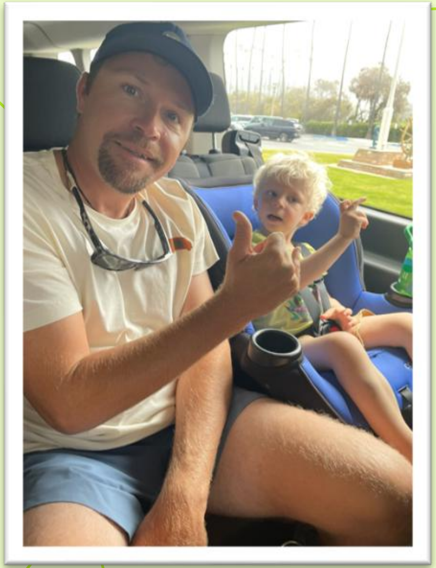
The diagnosis provided clarity

- **Kelly's brain works perfectly fine,** but he does not have the energy it needs to develop and function normally.
- We searched for years for the correct diagnosis. Kelly never 'fit' into autism or ADHD.
- Creatine Transporter Deficiency (CTD) explained a lot of Kelly's symptoms including:
  - Severe speech delay
  - Global developmental delay/intellectual disability
  - Autism-like features
  - Hyperactivity
- Those that suffer with CTD can also have seizures, failure-to-thrive, muscle weakness, GI issues, facial dysmorphia and many are non-verbal. Kelly does not have these symptoms but is at risk for developing them as the disease progresses.
- We are grateful know the diagnosis. We can now put all efforts into understanding and treating this disease.

The diagnosis was also devastating

- There is **no treatment or cure**.
- Kelly exhibits severe symptoms. While he is almost 5 years old, he is cognitively at the level of a 2.5-year-old. He is developing at 'half-speed' and continues to fall further behind during this **critical neurodevelopmental time**.
- It is unclear what Kelly will be capable of as he gets older. It is likely that he will live with us and need extensive care as an adult.
- As Kelly gets older, the benefit of a cure becomes less. We are racing against the clock.
- This is an ultra-rare disease, so awareness and funding are huge hurdles.

There is hope



There is an amazing non-profit

We are partnering with the Association of Creatine Deficiencies to raise money that will support research and development as well as spread awareness.

This is the website:

http://www.creatineinfo.org

There are pharmaceuticals

There are prospective drugs able to deliver creatine into the brain. These preform very well in pre-clinical trials, but we need funding to bring these to clinical trials. ACD is **partnering with drug companies and SO close to making this happen**. These medicines could potentially change the entire course of Kelly's life.

There will be gene therapy

Researchers across the globe are working to bring gene therapy to CTD as well as other ultra-rare genetic disorders. CTD is a single-gene mutation, making it prime for gene therapy. The ACD is partnering with Stanford, UCLA and Purdue amongst others to develop these advanced therapies for the future.

We need your help

Our Johnston Family goal is to raise 10% or $800K of the $8M needed. **Every dollar** will go directly towards funding research and development, including pharmaceutical clinical trials- starting as soon as **this summer.**

Kelly's Donation Link

Click hyperlink above or go to:
www.creatineinfo.org/kelly

OTHER WAYS TO GIVE

Set up a meeting to discuss our initiatives further:
Email our Executive Director, Heidi Wallis, at
heidi@creatineinfo.org

Mail check donations to:
ACD, 6965 El Camino Real, Suite 105-598,
Carlsbad, CA 92009

Donate with credit card, DAF, crypto, or bank transfer
online at: creatineinfo.org/make-a-donation

Donate stock:
Securities can be gifted to ACD via electronic transfer or by physically mailing the paper certificates to ACD. Your broker can contact heidi@creatineinfo.org to obtain the DTC number and further details.

Kelly III–and the family who loves him–thanks you!


RRS
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