MEDICAL HISTORY

THE

_____

FAMILY

MEDICAL HISTORY

IF LOST, PLEASE RETURN TO:

NAME: _____

PHONE #: _____

ADDRESS: _____

_____

_____

BASIC INFO

FULL NAME: _____

DATE OF BIRTH: _____ BLOOD TYPE: _____

EYE COLOR: _____ HAIR COLOR: _____

HEIGHT: _____ WEIGHT: _____

GENDER: _____ RACE/ETHNICITY: _____

MEDICAL NEEDS/CONDITIONS:	ALLERGIES:

PRIMARY CARE PHYSICIAN:

NAME: _____ PHONE #: _____

EMAIL: _____

ADDRESS: _____

_____

INSURANCE INFO:

POLICY HOLDER NAME: _____

INSURANCE NAME: _____

POLICY #: _____

GROUP #: _____

EMERGENCY CONTACTS

NAME: _____

PHONE: _____ RELATIONSHIP: _____

ADDRESS _____

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NAME: _____

PHONE: _____ RELATIONSHIP: _____

ADDRESS _____

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NAME: _____

PHONE: _____ RELATIONSHIP: _____

ADDRESS _____

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NAME: _____

PHONE: _____ RELATIONSHIP: _____

ADDRESS _____

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NAME: _____

PHONE: _____ RELATIONSHIP: _____

ADDRESS _____

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HEALTHCARE PROVIDERS

NAME: _____ EMAIL: _____

SPECIALTY: _____ PHONE #: _____

NURSES NAME: _____ FAX #: _____

ADDRESS: _____

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NOTES: _____

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NAME: _____ EMAIL: _____

SPECIALTY: _____ PHONE #: _____

NURSES NAME: _____ FAX #: _____

ADDRESS: _____

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NOTES: _____

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FAMILY MEDICAL HISTORY

MEDICAL CONDITION	NAME:	NAME:	NAME:	NAME:

CURRENT PRESCRIPTIONS

MEDICATION	DOSAGE	DATE FILLED	NOTES/SIDE EFFECTS

ALLERGY LOG

ALLERGY	NOTES	REQUIRED MEDICATION

MEDICATION LOG

DATE	TIME	MEDICATION/SUPPLEMENT	DOSE	NOTES

INJURY / ILLNESS HISTORY

DATE	DESCRIPTION	NOTES	REQUIRED MEDICATION

IMMUNIZATION RECORD

DATE	VACCINE	DOCTOR/CLINIC	NOTES/CHANGES

APPOINTMENTS

DATE: _____ QUESTIONS / NOTES: _____

TIME: _____ _____

APPT TYPE: _____ _____

DOCTOR: _____ _____

ADDRESS: _____ _____

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DATE: _____ QUESTIONS / NOTES: _____

TIME: _____ _____

APPT TYPE: _____ _____

DOCTOR: _____ _____

ADDRESS: _____ _____

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DATE: _____ QUESTIONS / NOTES: _____

TIME: _____ _____

APPT TYPE: _____ _____

DOCTOR: _____ _____

ADDRESS: _____ _____

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APPOINTMENTS

DATE: _____

TIME: _____

APPT TYPE: _____

DOCTOR: _____

ADDRESS: _____

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HEIGHT: _____

WEIGHT: _____

BLOOD PRESSURE: _____

TEMPERATURE: _____

HEART RATE: _____

RESPIRATION RATE: _____

OXYGEN: _____

OTHER: _____

QUESTIONS / NOTES: _____

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INSTRUCTIONS / PRESCRIPTIONS: _____

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THERAPY NOTES

DATE: _____

TIME: _____

DOCTOR: _____

THERAPY: _____

ADDRESS: _____

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QUESTIONS / NOTES: _____

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INSTRUCTIONS / PRESCRIPTIONS: _____

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HOSPITALIZATIONS

DATE: _____ DOCTOR(S) SEEN: _____

LOCATION: _____ _____

REASON: _____ DISCHARGED: _____

NOTES: _____ TREATMENT: _____

_____ _____

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_____ _____

_____ _____

DATE: _____ DOCTOR(S) SEEN: _____

LOCATION: _____ _____

REASON: _____ DISCHARGED: _____

NOTES: _____ TREATMENT: _____

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_____ _____

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_____ _____

_____ _____

ER / URGENT CARE

DATE: _____ DOCTOR(S) SEEN: _____

LOCATION: _____ _____

REASON: _____ DISCHARGED: _____

NOTES: _____ TREATMENT: _____

_____ _____

_____ _____

_____ _____

_____ _____

_____ _____

DATE: _____ DOCTOR(S) SEEN: _____

LOCATION: _____ _____

REASON: _____ DISCHARGED: _____

NOTES: _____ TREATMENT: _____

_____ _____

_____ _____

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_____ _____

SURGICAL HISTORY

DATE: _____ PROCEDURE: _____

LOCATION: _____ PHYSICIAN: _____

REASON: _____

POST-SURGERY NOTES: _____

DATE: _____ PROCEDURE: _____

LOCATION: _____ PHYSICIAN: _____

REASON: _____

POST-SURGERY NOTES: _____

LAB WORK

DATE: _____

LAB WORK: _____

RESULTS/NOTES: _____

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DATE: _____

LAB WORK: _____

RESULTS/NOTES: _____

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RESULTS/NOTES: _____

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BLOOD PRESSURE LOG

DATE	TIME	BLOOD PRESSURE	HEART RATE	NOTES

BLOOD SUGAR LOG

DATE	TIME	LEVEL	NOTES

DENTAL LOG

DATE	DENTIST	PROCEDURE/REASON	NOTES

DENTAL VISITS

DATE: _____ DENTIST: _____

TIME: _____ LOCATION: _____

REASON: _____ FOLLOW UP APPT: _____

CLEANING: Y / N RESULTS/COMMENTS: _____

X-RAY'S: Y / N RESULTS/COMMENTS: _____

PROCEDURES: _____

_____

NOTES: _____

_____

DATE: _____ DENTIST: _____

TIME: _____ LOCATION: _____

REASON: _____ FOLLOW UP APPT: _____

CLEANING: Y / N RESULTS/COMMENTS: _____

X-RAY'S: Y / N RESULTS/COMMENTS: _____

PROCEDURES: _____

_____

NOTES: _____

_____

DENTAL HISTORY

DATE: _____ PROCEDURE: _____

LOCATION: _____ DENTIST: _____

REASON: _____

AFTER VISIT NOTES: _____

DATE: _____ PROCEDURE: _____

LOCATION: _____ DENTIST: _____

REASON: _____

AFTER VISIT NOTES: _____

EYE CARE

DATE: _____ NOTES: _____

TIME: _____ _____

DOCTOR: _____ _____

RIGHT EYE: _____ _____

LEFT EYE: _____ _____

COST: _____ _____

DATE: _____ NOTES: _____

TIME: _____ _____

DOCTOR: _____ _____

RIGHT EYE: _____ _____

LEFT EYE: _____ _____

COST: _____ _____

DATE: _____ NOTES: _____

TIME: _____ _____

DOCTOR: _____ _____

RIGHT EYE: _____ _____

LEFT EYE: _____ _____

COST: _____ _____

MEDICAL EXPENSES

DATE	DESCRIPTION	BILLING PARTY	TOTAL BILLED	INS. PAID	DEDUCT AMNT.	CO-PAY AMNT.	OUT OF POCKET

WEEKLY SCHEDULE

MONDAY	TUESDAY	WEDNESDAY	THURSDAY

FRIDAY	SATURDAY	SUNDAY	NOTES:

NOTES

NOTES

CHECKLIST

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CHECKLIST

Types of CHAMP1 Gene Mutations Explained

Missense Mutation
Nonsense and Frameshift Mutations
Microdeletion

Missense Mutation

A missense mutation occurs when there is a mistake in the DNA code and one of the DNA base pairs is changed (example, A is swapped for C.) This single change means that the DNA now encodes for a different amino acid, known as a substitution.

Children with CHAMP1 missense mutations are believed to have the full CHAMP1 protein in a mutated form.

Diagnostic: Whole Exome Sequencing, Autism Panel, Epilepsy Panel, ID Panel

Mutations Resulting in a Stop Codon

Nonsense Mutations occur when there is a mistake in the DNA code and one of the DNA base pairs is changed. This single change can cause a set of three nucleotides (a codon) to now encode a stop codon, instead of an amino acid. This change in the DNA and messenger RNA (mRNA) will signal protein synthesis to stop before the whole protein is made.

Frameshift Mutations occur when nucleotide bases are inserted/deleted, and the number of nucleotide bases inserted/deleted is not divisible by 3. This event causes a change in the reading frame, since a protein is created by reading sets of 3 nucleotides at a time to determine which amino acid is encoded by the sequence. Frameshift mutations may also result in an early stop codon, similar to nonsense mutations.

Patients with CHAMP1 mutations that have stop codons are confirmed to have around 50% of the CHAMP1 protein needed and a more severe phenotype.

Diagnostic: Whole Exome Sequencing, Autism Panel, Epilepsy Panel, ID Panel

Microdeletion

A loss of a piece of chromosome.

Patients with a microdeletion might be missing 1 copy of the CHAMP1 gene entirely. They may also be missing other genes on the 13th chromosome in addition to CHAMP1.

Diagnostic: Chromosomal Microarray

CHAMP1 Gene Mutation

COMMON QUESTIONS

1 WHAT IS CHAMP1?

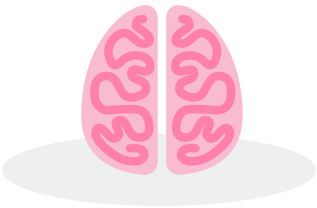
Short for Chromosome Alignment- Maintaining Phosphoprotein. CHAMP1 an ultra rare genetic disease discovered in 2015. It is located on chromosome 13 and is said to be crucial in cell division called mitosis. The mutation of the CHAMP1 gene is referred to as haploinsufficiency, this means out of the 2 alleles the mutation causes a loss of function to one leaving those affected with a single functional copy. Patients have a reduction in the CHAMP1 protein below the 50% which is necessary for proper neurological function and development.

2 WHAT CAUSES A CHAMP1 MUTATION?

CHAMP1 is most commonly a de novo mutation, meaning it is not inherited from the parents but completely new to the genome of the individual. There are also reported cases of missense mutations inheritably passed down. These patients express a milder phenotype.

3 WHAT ARE THE SYMPTOMS?

Children with CHAMP1 have varying levels of functionality. The lack of protein is responsible for a multitude of symptoms. The most recurrent symptoms of significance include global developmental delay/Intellectual disability and severe speech delay/loss. Others: Developmental delay, severe speech delay/loss of speech, autistic traits, feeding difficulties from birth, reflux, abnormal MRI (white matter, microcephaly, structural deformities), hypotonia, epilepsy, vision issues, incontinent, anxiety, sleep issues, flat nose bridge, low set ears, small stature, CVS(Cyclical vomiting Syndrome) Dental issues (small teeth, misaligned, Baby teeth stay in to long.)



4 HOW MANY PEOPLE ARE DIAGNOSED?

Currently there are approximately 100 patients diagnosed with a CHAMP1 mutation in the world. There are individuals with CHAMP1 in over 21 countries with boys and girls affected alike.

5 IS THERE A CURE OR TREATMENT?

Currently there is no cure or significant treatment for CHAMP1 yet but early and personalized intervention is recommended and necessary to thrive. Traditional therapies have been shown to show some improvement over time.

Get Involved! Here's how...

Below is a list of important things you can do to get involved. Each item makes such a difference in the lives of our CHAMPS! We're so excited to have you on this journey with us.

1 PROVIDE A SKIN SAMPLE

Corielle Institute is collecting skin samples from our CHAMPS. This will program the sample into a fibroblast which can be studied, and/or turned into a IPSC (induced pluripotent stem cell) line. You can donate blood too, but skin is the priority and it would be more important to get blood to CHOP first (see below.)
For more information: https://www.youtube.com/watch?v=3hLr8ZNb87c&t=10s

2 DONATE BLOOD

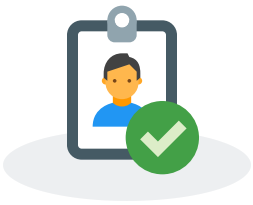
Donate a blood sample to Children's Hospital of Philadelphia (CHOP.) Your child's specific mutation will be researched to identify ways to help find treatments for our children.
Watch how here: https://www.youtube.com/watch?v=HFZxKfBcpyQ&t=47s

3 HELP DR. RICHARD KELLEY

Currently we know there are amino acid abnormalities found in CHAMP1 patients. Dr. Richard Kelley is collecting blood work to study and look for a possible treatment of this metabolic disturbance. **Tests include**: Amino acids (plasma), lipid panel (cholesterol fractions + triglycerides), comprehensive metabolic panel, lactic acid (plasma), vit. E level, & total homocysteine level. Your Pediatrician will be able to assist you in obtaining these specific lab tests.
Email Dr. Richard Kelley directly at: rikelleymd@comcast.net

4 SIMONS SEARCHLIGHT

You can also do our existing Natural History Study through Simons Searchlight. Visit SimonsSearchlight.org, register and upload your genetic report. They will reach out to you to set up a phone call!
Current supported languages include: English, Dutch, French, and Spanish
For further questions, email: coordinator@simonssearchlight.org



5 FINANCIAL SUPPORT

Please consider making a donation if possible. We would also love to come along side you and help create an online fundraiser when you're ready! Lastly, videos like this help raise awareness while sharing our incredible stories.
Watch here: https://youtu.be/C--NTSi0GIs

CHAMP1 RESEARCH FOUNDATION

Follow us on social media for updates and information! Be sure to like and share our posts to raise further awareness for our CHAMP1ons.

CHAMP1 Research Foundation

@champ1research