8p Hero Passport

Name:

Date of birth:

Sex: Female

Genetic Diagnosis

Date of testing: NA

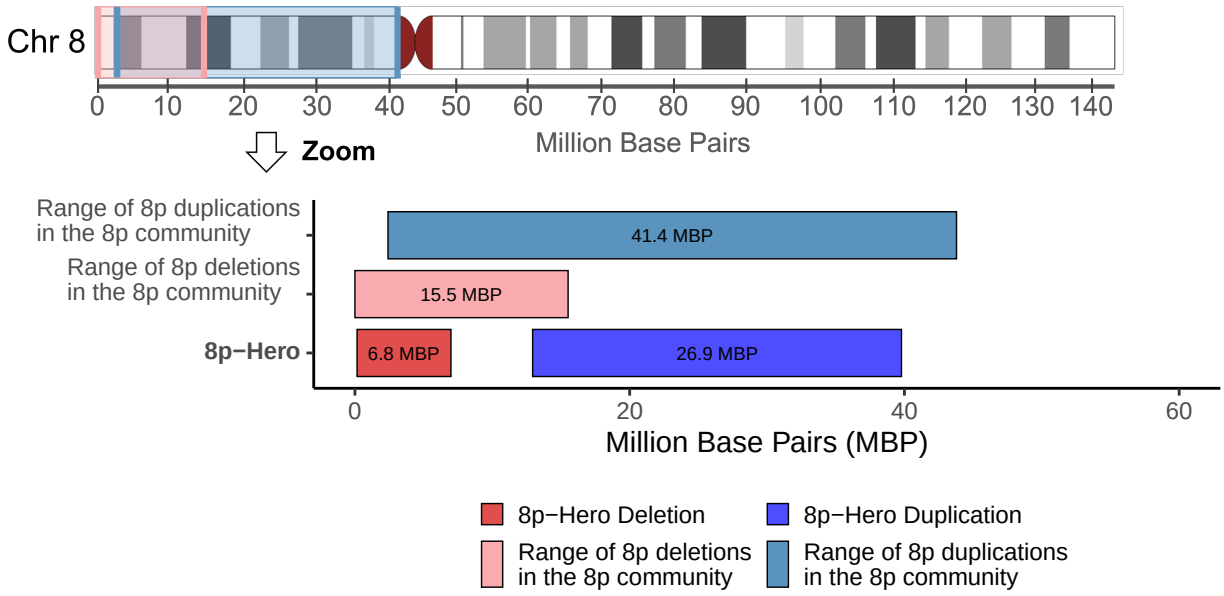
Test type: Other

Genetic finding: Chromosome 8p inverted duplication deletion (8p deletion: 8p23.3 (155,048) to 8p23.1 (6,989,234); 8p duplication: 8p23.1 (12,934,123) to 8p11.1 (39,786,863))

Duplication size: 26,852,740 BP. Median size among other 8p heroes: 24,194,174 BP.

Deletion size: 6,834,186 BP. Median size among other 8p heroes: 6,837,807 BP.

Inheritance: Parental testing not done



Family Genetic Testing Recommendation

Given the genetic nature of chromosome 8p disorders, we recommend that family members consider genetic testing to better understand potential hereditary patterns. Family genetic testing can provide valuable insights into the unique genetic makeup of each family member, helping clinicians tailor care more effectively and enabling families to make informed decisions about health and wellness.

How you can connect with my 8p Hero

When interacting with my 8p Hero, it's important to approach with warmth, patience, and respect. Here are a few ways you can create a meaningful connection:

1. **Start with a greeting:** Say hello clearly and make eye contact when possible. Using my Hero's name helps them recognize your friendly intent.
2. **Pause for a response:** After asking a question or giving an instruction, wait for at least 3 seconds. My Hero may need extra time to process and respond, which could be in the form of a gesture, a nod, or even an eye blink.
3. **Use open body language:** Keep your gestures expressive and welcoming. A warm demeanor often encourages my Hero to feel comfortable in your presence.
4. **Respect personal space but be open to connection:** If my Hero shows interest in a hug or physical touch, depending on your comfort level, it's okay to offer a hand or a gentle hug.
5. **Other ways to connect to my 8p hero:**

Chromosome 8p Disorder Summary

Chromosome 8p disorder is a rare genetic condition that is known to affect 550 individuals worldwide, but has a reported prevalence of 1:10,000 - 1:30,000. Chromosome 8p disorder is caused when there is a rearrangement of genetic information before birth, on the short arm (the p arm) of the 8th chromosome. This genetic rearrangement can be the deletion of information, duplication of information, or inverted duplication and deletion (invdupdel). Chromosome 8p disorder has systemic effects, meaning it impacts cells and tissues throughout the body, rather than being confined to a single organ. The chromosomal rearrangement typically arises from spontaneous (de novo) changes early during embryonic development, for reasons that remain unclear. The severity and specific features of 8p disorders can vary significantly based on the amount of genetic information that has changed in addition to other factors that we are still working to understand.

There are several types of therapies that can be effective for children with 8p disorders, depending on their symptoms, these include speech therapy, occupational therapy, physical therapy, vision therapy, ABA therapy, aquatic therapy, feeding therapy, hippotherapy as well as assistive and augmentative communication devices. In the absence of treatments for chromosome 8p disorders, clinicians at Project 8p's multi-disciplinary clinic recommend treating symptoms with standard clinical practice as they arise.

Unfortunately, no targeted therapies for chromosome 8p disorders exist yet. As such, current treatment focuses on managing symptoms as they appear and implementing 8p standards of care. Project 8p is dedicated to accelerating treatments for tomorrow. We can facilitate collaboration with our clinical experts to support your care plan and offer tailored resources. For further assistance, contact our clinical team at engagement@project8p.org.

Current Medication & Supplement Information

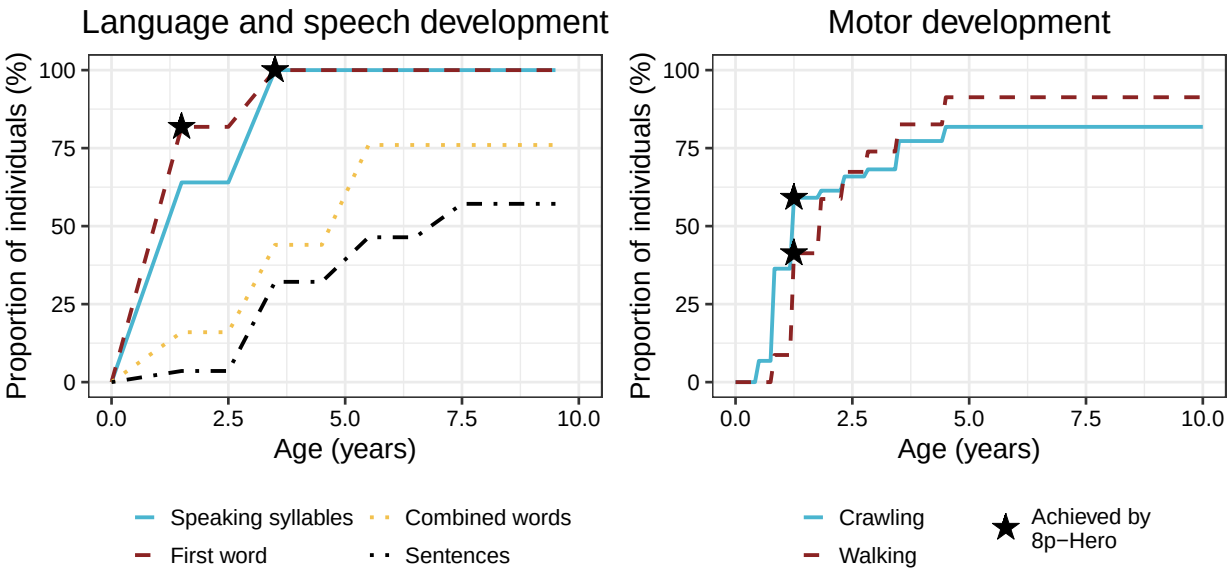
Medication	Frequency	Dosage
Cetirizine	daily	5ml
Magnesium sulfate	daily	1/8 tsp
THC oils	a.m./bedtime	NA
Supplement	**Frequency**	**Dosage**
Benefiber	daily	variable
GBX	daily	Amare

Prior medications can be obtained from the Appendix.

Prenatal and Birth History

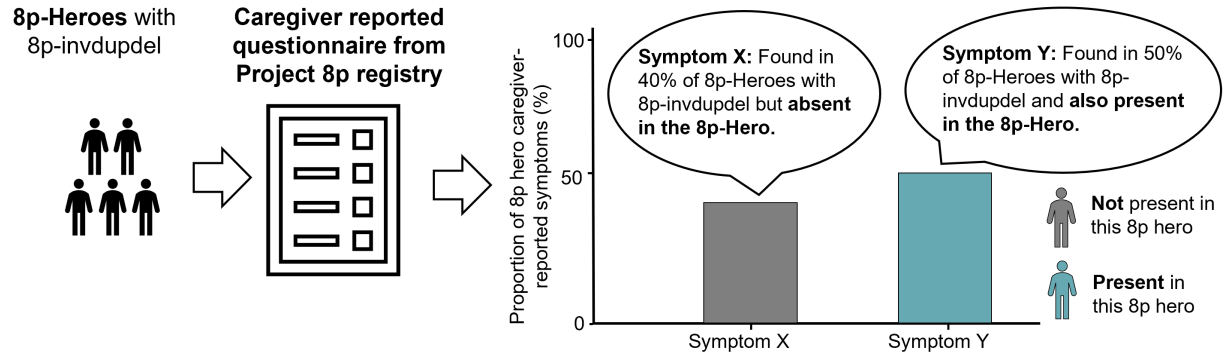
General information	Your 8p Hero	Other 8p Heroes with 8p inverted duplication/deletion median (25-75% quantiles)
Weight	Unknown	-
Length	45 cm 17.7 inch	48.3 cm (47 cm - 51.4 cm) 19 inch (18.5 inch - 20.2 inch)
Head Circumference	Survey incomplete	33 cm (31.8 cm - 34.1 cm) 13 inch (12.5 inch - 13.4 inch)
Gestational	32 - 36 weeks	-
Prenatal & birth complications		
NICU Admission	Yes	53.3%
NICU Reason	Breathing Difficulties	-
Days at the Hospital following NICU Admission	2	4 (2 - 10.75)

Developmental Milestones



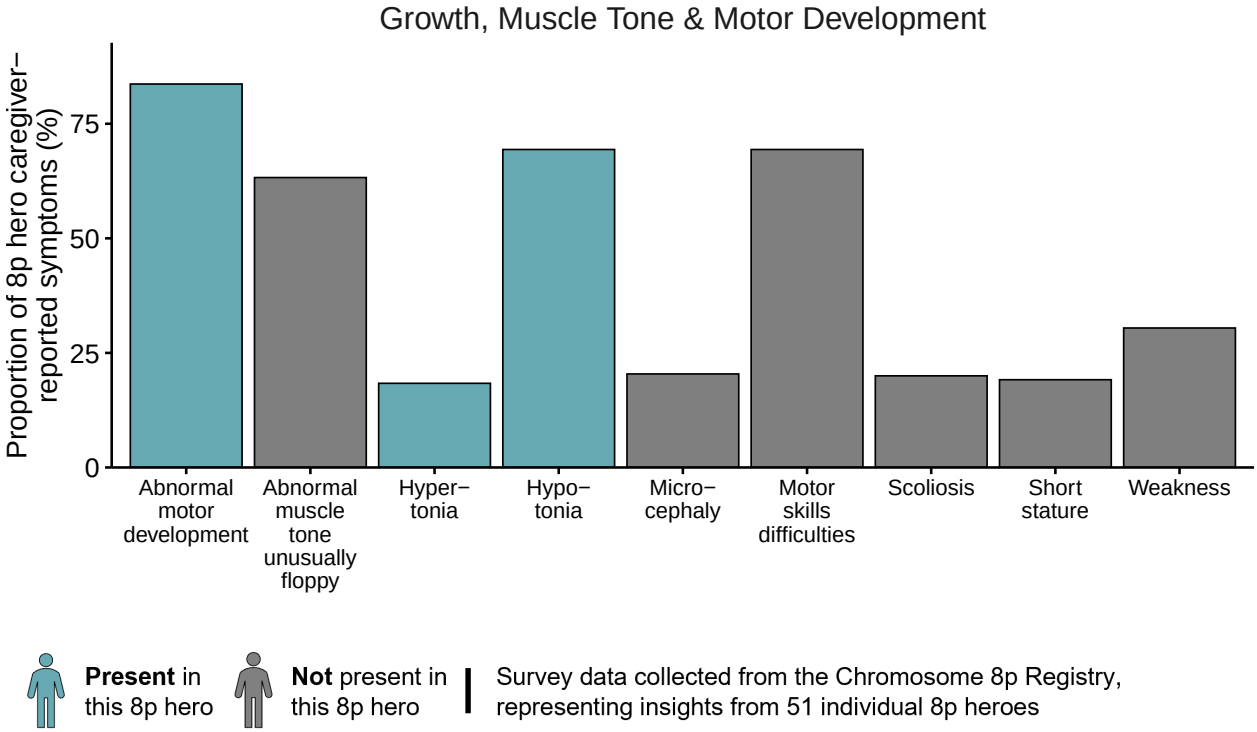
**For developmental milestones achieved, the 8p hero comparison shown is the percentage of 8p heroes that have achieved that milestone by the same age as your 8p hero. If the age when the 8p hero achieved the milestone is known it is indicated by a star.*

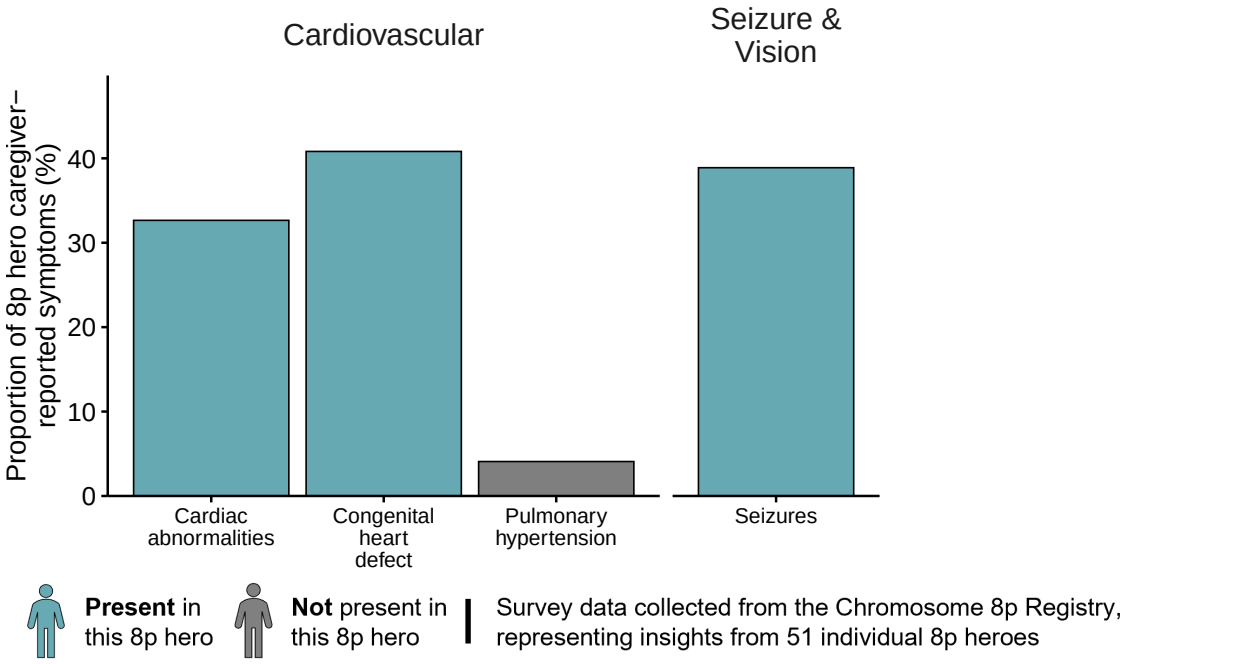
How to interpret symptom frequency plots



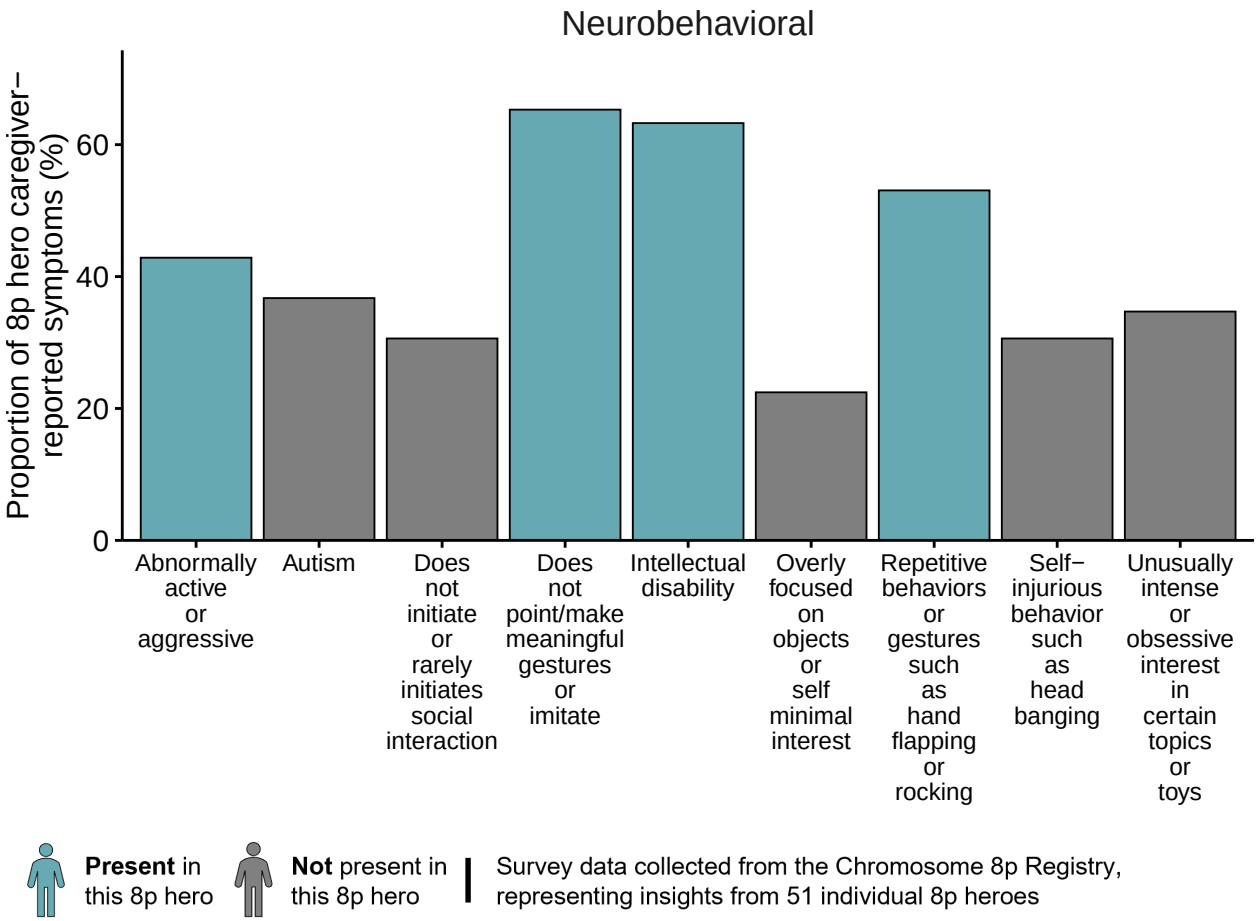
The age of assessment of the 8 hero was 6 year(s) and 5 month(s). Please note that the comparison group includes all 8p heroes with 8p inverted duplication/deletions. Be aware that some 8p heroes may not display a clinical characteristic at the time of assessment but might develop it later. The fraction of 8p heroes with a clinical characteristic might thus be underreported.

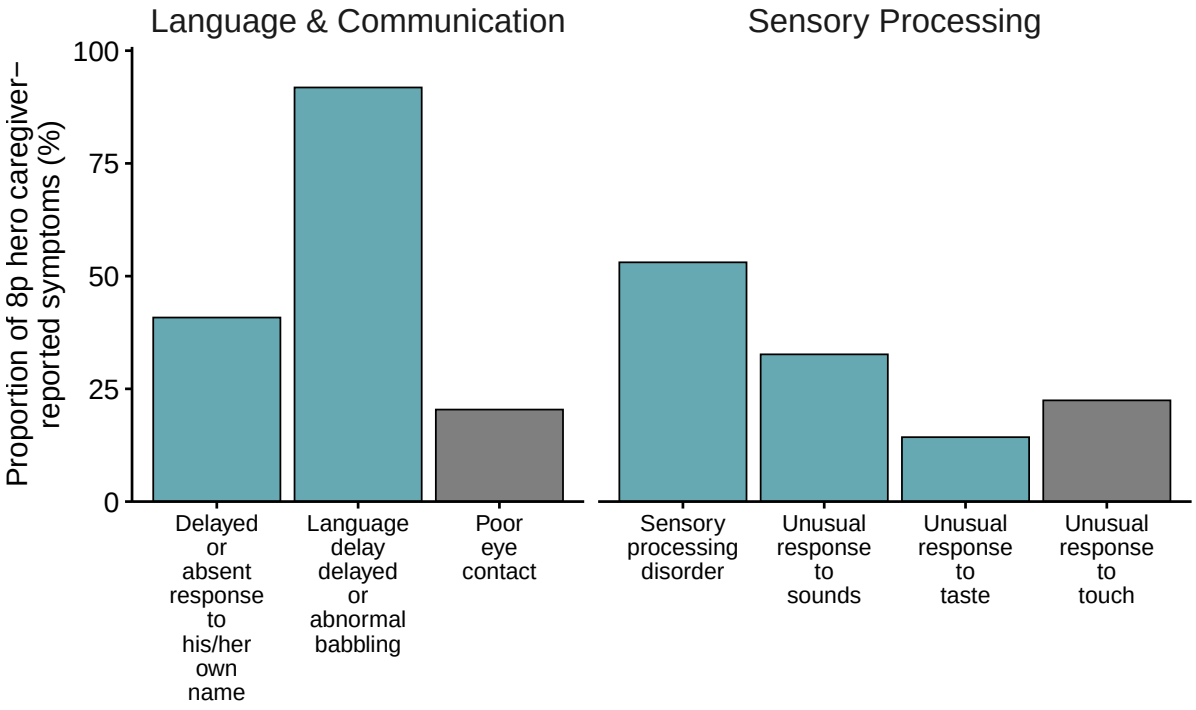
Physical Health



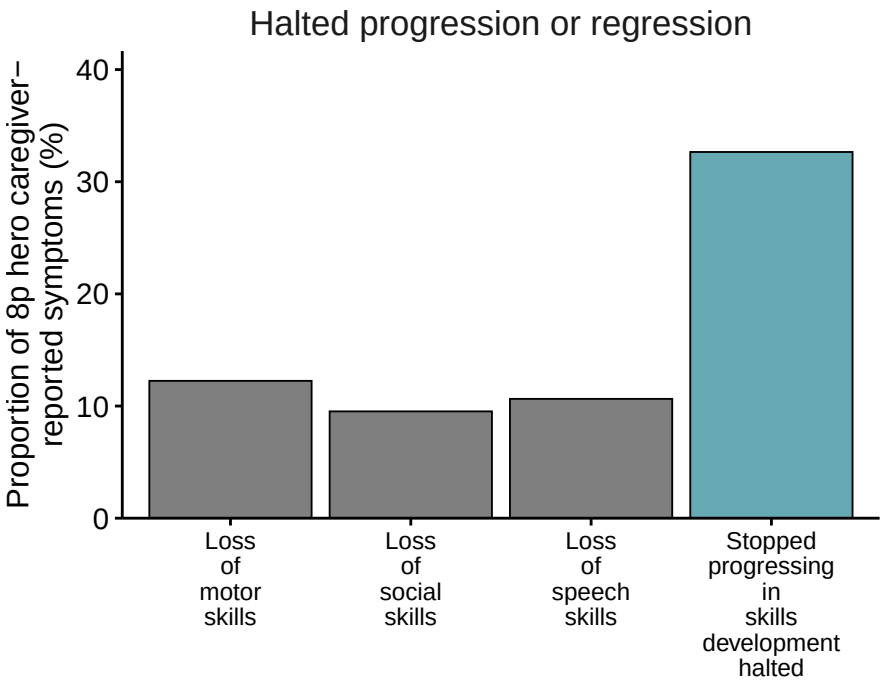


Behavioral and Cognitive Development





Present in this 8p hero **Not** present in this 8p hero | Survey data collected from the Chromosome 8p Registry, representing insights from 51 individual 8p heroes



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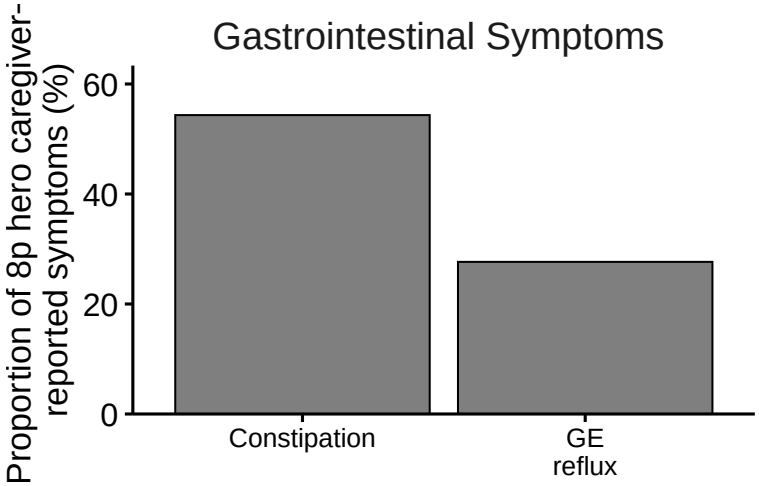
Gastrointestinal Health



Present in this 8p hero | **Not** present in this 8p hero | Survey data collected from the Chromosome 8p Registry, representing insights from 51 individual 8p heroes

The following Gastrointestinal health symptoms have been documented in the 8p population but we have insufficient data to provide prevalence estimates at this time: Diarrhea, jaundice, abdominal pain, enlarged liver, malabsorption, liver function abnormalities, gallbladder disease, vomiting, cyclic vomiting, inflammatory bowel disease, colic

If you have any questions about any of the information provided in this 8p Hero Passport, or would like to update any of your 8p hero's information, please contact us at engagement@project8p.org.

Visit the 8p Patient Navigator for care guidelines and recommendations: https://project8p.org/.

Join the cause! (Find more details here: https://project8p.org/join-the-cause/)

Project 8p Foundation is a registered 501(c)(3) charitable organization with EIN 83-2545342. All contributions are deemed tax-deductible.

Contact Information

info@project8p.org

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Appendix

Prior medication and supplements not continued

Medication	Frequency	Dosage	Stop Reason
None reported	NA	NA	NA
Supplement	**Frequency**	**Dosage**	**Stop Reason**
Vitamine C	weekly	NA	See notes