TOGETHER TOWARDS TREATMENT

Project 8p Foundation

5 Year Strategic Plan

2021
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Section I: Five Year Strategic Plan

The Project 8p Foundation is a non-profit 501(c)(3) corporation that means business.

Vision: Cure genetic brain disorders with novel technologies that are accessible to all.

Who we serve

8p heroes are individuals with a chromosome 8p disorder who have deleted and/or duplicated genes that affect their brains and hearts. They live with everyday challenges that most of us take for granted, like walking, talking, and just being healthy. We have identified over 500 people but research estimates indicate there are over 500,000 all over the world.

Our Community: 8p heroes, caregivers, families, providers, therapists, clinicians, researchers, funders, investors, sponsors, industry partners, and general supporters.

Our disorder knows no differences.

Our families are all over the world from small rural towns of varying socioeconomic statuses, racial and religious backgrounds, ethnicities, nationalities, ages, gender, sexual orientation, and diverse lifestyles and experiences. Diversity benefits us with new perspectives at all levels of the organization. We are socially responsible as access to Project 8p’s initiatives are for our entire community and free from bias. Our organization will be representative of the diverse population we serve. We will engage in partnerships that align with our social commitment and core values.

Our why

Having an 8p diagnosis was a black hole. 8p heroes deserve our energy for life-changing treatments.

Mission: to empower a unified community for chromosome 8p heroes for a meaningful life today while accelerating treatments for tomorrow.

Our five year strategic plan with priority goals and actions will allow us to reverse the challenges that face us:

- We are in a race against time. Time is Brain. Everyday as time passes, the critical window of intervention to support brain development and mental health gets compromised.
- There are no treatments
- There are no expert doctors to improve our understanding to manage care or answer what to expect in prognosis or what symptoms/disorders we can prevent in a lifetime if we knew more
● There is no current roadmap for scientists or clinicians or families
● There is a huge unmet need in rare and orphan diseases with limited resources.
● Isolated families are in the dark with no way to connect to other families meaningfully

When parents first receive the news that their child has been diagnosed with a rare chromosome condition, most feel scared, lonely, confused, and devastated. Prior to the formation of Project 8p, families searching for answers yielded little results with almost nothing known about the diagnosis and their future.

There is a need to bridge the gap of knowledge and connect information and people. A need to harness the power of more than 1 child to many others whom we can treat. Therefore, our mantra is “Together Towards Treatment.”

What we do

Project 8p Foundation was started in November 2018 to connect the missing clues and provide answers and solutions. Project 8p provides hope for families to have a positive life altering resource at the time of diagnosis and growth.

We are creating a patient-led community to help patients and caregivers have resources to improve a loved one’s health outcomes and to become active participants and advocates for translational research to find treatments. The possibilities of genomic medicine are only achievable with a strong community who advocates together in numbers and in turn can influence and generate involvement from scientists, researchers, biotech, pharma, and healthcare industries.

In the last several years with resilience, we initiated strategic projects in science and medicine. With these Key Opinion Leaders, we have validated that the Time is Now to Invest with Us.
Our Impact: Priority Goals

Constant innovation and creativity are required to solve our puzzle. We prioritized 4 key goals. Moreover, we need to build capacity to grow our operations. The 4 goals are interdependent and will inform our decisions and overall impact.

The Paradigm Shift

What if 8p was a clue to the puzzle of common disorders? Our organization is a team of detectives. The genes located on chromosome 8p, missing or duplicated are linked to Alzheimers, Parkinsons, cancer, autism, schizophrenia, and more common disorders.

What if we knew what to expect when 8p heroes grow older and we can prevent new unknown symptoms? Do their lives become more difficult?

Project 8p is championing the view that if the causes of common diseases are unique for every person, then every individual with a rare disease is an important clue to solving the puzzle and a gift to science. Designer drugs, precision medicine, gene therapies are all relevant to this problem. The field of precision medicine takes this view that the causes of common disorders differ from person to person, arising from a cocktail of genetics, environment, lifestyle, nutrition, and more. If we push this view to its limit, every individual with some disease is seen to have an ultra-rare version of the disorder that is uniquely their own. If we move away from curing a disease to curing a human being, then aren’t we all rare as individuals? Rare diseases become obsolete, orphan diseases find a home, and Project 8p will achieve its vision.
## Impact Summary of Goals: Five-Year (2021-2026)  

<table>
<thead>
<tr>
<th>GOALS</th>
<th>AREAS OF FOCUS</th>
<th>MEASURE OF SUCCESS (KPI)</th>
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<tbody>
<tr>
<td><strong>1. Fund sponsored research to support innovations and identify therapeutic targets</strong>&lt;br&gt;2. Build a robust data platform with the Chromosome 8p Registry to leverage knowledge into meaningful outcomes for 8p heroes&lt;br&gt;3. Partner with established and emerging Key Opinion Leaders (“KOLs”)</td>
<td>** ● Fund 6 sponsored research projects and 6 IND Enabling studies (including non-academic) <strong>&lt;br&gt;</strong> ● Clinical trials in 2 therapies <strong>&lt;br&gt;</strong> ● Increase participants into the Data Platform and Biobank by 100% annually <strong>&lt;br&gt;</strong> ● Strategically partner with 15 organizations and collaborate with 85 KOLs in neuroscience and chromosomal disorders **</td>
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<td><strong>1. Build evidence to define high quality of care for 8p heroes</strong>&lt;br&gt;2. Manage and maintain a scalable Neurogenetics Multidisciplinary Clinical Research Network&lt;br&gt;3. Gather intelligence from families to inform patient needs and research priorities</td>
<td>** ● 5 Clinical Centers (with at least 2 international) <strong>&lt;br&gt;</strong> ● Increase the number of 8p heroes attending clinics by &gt;20% annually <strong>&lt;br&gt;</strong> ● Publish standard of care for 8p heroes <strong>&lt;br&gt;</strong> ● Outreach to 100% of families affected **</td>
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<td><strong>1. Solidify diversified and sustainable funding sources</strong>&lt;br&gt;2. Educate our investors about their value in our collective global impact&lt;br&gt;3. Create community activation partnerships</td>
<td>** ● Increase donor and volunteer support to exceed $5,000,000 <strong>&lt;br&gt;</strong> ● Build master prospect list targeting 100 family foundations and 2,000 individual and major donors **</td>
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<td><strong>1. Provide education and advocacy tools to the patient community</strong>&lt;br&gt;2. Significantly increase and maintain engagement with a user experience (“UX”) that transforms into a sustainable patient-led and patient-centered platform&lt;br&gt;3. Define content strategies for the patient community that connect to and influence the broader community</td>
<td>** ● Patient Leadership Board to oversee family engagement across all goals as the voice of 8p families <strong>&lt;br&gt;</strong> ● Host bi-annual conferences and informal retreats annually; award travel scholarships <strong>&lt;br&gt;</strong> ● Increase activity on the data platform with 100% compliance on survey completion <strong>&lt;br&gt;</strong> ● Grow social media followership by 100% annually **</td>
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1More detail available upon request
Milestones Achieved

- March 2021 – Virtual meeting for Commission on Novel Technologies for Neurodevelopmental Copy Number Variants
- Fox NY TV News Feature Story as Best of the Big Idea of 2019
- July 2021, Colorado - Joint Rare Disease Moving Mountains Science and Family Conference
- Declared August 8th to be recognized as Chromosome 8p Day (8/8)
- Funded and published natural history study with Dr. Wendy Chung in Genetics in Medicine: Clinical and genomic characterization of 8p cytogenomic disorders
- Awarded CZI Rare as One Grant ($465,000) in February 2020
- Established a Commission on Novel Technologies for Neurodevelopmental CNVs to emphasize the importance and burden of Chromosome Disorders like 8p.
  - Uniquely patient-led with signed charters by committed researchers for transparent team science approach
- Obtained an approved IRB supporting to launch the Chromosome 8p Registry (IRB Protocol #20203404)
- Setup the first 8p Biorepository with specimens and stem cells of affected families
- Launched 1st Neurogenetics Multi-Disciplinary Clinic at Children’s Hospital of Colorado
- Founder as a finalist in 2021 Loreal Women of Worth
Core Values

**WE DISRUPT**

WE CHALLENGE THE STATUS QUO FOR SCALABLE IMPACT

**WE RACE**

WE RACE AGAINST TIME AND TIME IS BRAIN

**WE PERSEVERE**

WE ARE RESILIENT - INSPIRED BY 8P HEROES WHO NEVER STOP PERSEVERING

**WE CONNECT**

WE BRIDGE DIVERSE HEROES TO ONE COMMUNITY THAT STANDS FOR EQUITY IN HEALTH AND SCIENCE

The Future

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**FIRST CHROMOSOME THERAPY**

Today in 2025, this child can run in the park with friends and give a tight hug to those that made the difference. For the first time, 8-year old Cameron can say the words "thank you for changing my life."

First Chromosome Therapy significantly improves motor function, cognition, speech, and survival.

In 2018, 8p disorders were poorly understood with no active researchers and no expert clinicians.

Patients led the path to treatment by uniting stakeholders in their perseverance "Together towards treatment"
Section II: The Strategic Plan in Detail

About Chromosome 8p Disorders

Chromosome 8p is a rare genetic condition with approximately 350 to 500 patients around the world and counting. About 4.5 to 5 percent of a human’s total DNA is located on chromosome 8p – about 700 to 1,400 genes. Chromosomal conditions typically impact every cell in the body, so patients are affected systematically and often require significant support.

The impacts of these 8p conditions vary between individuals and their abilities. Some people may not learn to talk, may need to use a wheelchair, and may need more support services. Others are mobile, can communicate, and can achieve greater independence. The majority of 8p individuals have moderate to severe developmental delays including disordered acquisition of cognitive and social competence and delays in reaching developmental milestones. These patients average 1,500 hours of therapy a year and at least one doctor visit a month. The disorder can result in:

- Global Developmental Delays
- Intellectual disabilities
- Seizures (epilepsy)
- Autism
- Apraxia (difficulty performing and coordinating movements i.e. feeding oral motor, speech communication)
- Sensory processing disorder
- Motor planning
- Vision impairments
- Growth deficiency
- Feeding and digestive challenges
- High (hypertonia) and low (hypotonia) muscle tones
- Musculoskeletal issues such as hypermobility (lax joints)
- Mild craniofacial differences
- Sleep disorders

Scope of the Bigger Problem

Rare Diseases are a Minority, but Collectively we are Critical to Solve Disease

Did you know that 1 out of 10 people you know will have a rare disease in their lifetime? (U.S. genetic poll)

Rare diseases are anything but rare: as many as 7,000 rare diseases affect approximately 400 million people worldwide. Yet, for an individual patient and their loved ones, living with a rare
disease can be isolating. The diagnostic odyssey averages a daunting 7 years as patients go from doctor to doctor, searching for answers. And obtaining an accurate diagnosis — while critically important — seldom points to a clear actionable plan for treatment.

Why are there so few treatments for rare diseases? A major reason is that the underlying biology of most of these diseases is poorly understood — Bottom line: it’s difficult to treat any disease if you don’t understand its biology. The complex ecosystem of expertise required divides an already small space, particularly with rare diseases that involve large copy number variants like chromosome 8p. The silver lining is there is quite a bit of overlap across rare diseases and common diseases and potential economies of scale.

Expanding the 8p Reach

We are connecting datasets existing in silos with an investment in advanced bioinformatic tools to help us get meaningful insights into the disease, how it manifests, and potential therapeutic targets.

*The cost of the Human Genome Project in 2013 came down from $1.3 Billion to less than $1,000 to sequence a human being.*

The upside is that pivotal clinical trial studies of orphan drugs for rare neurological diseases are approved with few trials and with a smaller number of participants with the support of the Orphan Drug Act, offering incentives for biopharma development in orphan diseases. Therefore, today, we are primed with the tools and industry support to accelerate research with similar cutting-edge technology to help us find treatments and therapies for brain disorders.

Project 8p has sought to strategically broaden the tent to unite stakeholders and resources to focus on chromosome disorders collectively. One such common genetic similarity is with Down Syndrome which involves duplications or three copies of chromosome 21 presenting with neurological and other challenges. In 2020, we launched a [Commission on Novel Technologies for Neurodevelopmental Copy Number Variants (CNV)](https://www.chanzuckerberginitiative.org/8p) that affect the brain. This effort has been seed funded by the [Chan Zuckerberg Initiative](https://www.chanzuckerberginitiative.org/). If we continue to behave as risk-takers pushing the envelope with creativity, then we believe that cross-disorder research and basket trials will likely get us to treatments faster by leveraging economies of scale alongside other disorders beyond 8p.

To date, we have partnered with key opinion leaders (KOLs) that have committed to actively participating in this approach. If we are successful in leading novel approaches for chromosome disorders, Project 8p will serve as a thought leader in the field of neurologically impacted chromosome disorders and simultaneously advance the prioritization of 8p disorders.
As we look to the future and the challenges facing families with chromosome 8p, we need to ensure that new and innovative technologies are being developed to better understand 8p and guide research, diagnosis, and treatment.

Our Commitment

Over the next five years, Project 8p will grow our capacity requiring a significant investment to enable deeper engagement in the issues most important to the 8p community. By serving our 8p community at all times, we will remain accountable for our decisions towards the strategic goals. This will support a sustainable organization with the expertise to better traverse the challenging terrain ahead.

We will utilize advanced technology in all aspects in pursuit of our goals. We will continue to partner with other organizations that share our goals and vision.

Finally, we want to create a culture that resonates with our core values across the organization with people that are proactive and celebrate new perspectives, risk-taking, and unconventional approaches for the greater good.

Goal: Accelerate Research for 8p Related Disorders

Focus Areas:

1. Fund sponsored research to support innovation and identify therapeutic targets

2. Build a robust data platform with the Chromosome 8p Registry and Biorepository to leverage knowledge and materials into meaningful outcomes for 8p heroes
3. Partner with established and emerging Key Opinion Leaders ("KOLs")

With a personalized and optimized treatment plan being the future state, we must equip patients and families with tools to navigate all areas of diagnosis and prognosis easily and cost-effectively. This includes daily management, medical and therapeutic intervention, diet and nutrition, education, and socialization to cultivate a knowledgeable community. With this exchange of information, we can better understand the disease to find a treatment.

Currently, our investment in basic science research is critical to establish the genetic underpinnings and biological mechanisms for potential therapeutic targets for treatment. Project 8p Foundation will lead investing in research projects with non profit academic institutions as well as companies and contract organizations. One major program we have launched is the a patient-led Commission on Novel Technologies for Neurodevelopmental Copy Number Variants (CNVs). By attracting funders and the scientific community broadly about disorders like Chromosome 8p and the scale of the importance of the public health burden, we will position ourselves as a critical disease. We are currently sponsoring studies to address pressing research questions and provide the research and medical community with free exchange of data and biospecimens.

To support this effort, Project 8p has built and launched an international Chromosome 8p Registry as well as a biorepository of family biospecimens. The Chromosome 8p Registry will serve as a retrospective and prospective natural history study that connects patient-reported outcomes, clinician reported data, and researcher reported data. In addition, it will link relevant information to the biospecimens for research but also report back to families with transparency about their participation in research.

The Registry and biorepository platform will take advantage of the latest technology and advances in patient registry development. We envision a dynamic and responsive resource that will constantly incorporate the latest disease specific information and research developments personalized and relevant to each participant.

The Chromosome 8p Registry is GDPR and HIPAA-compliant and will be used to collect and store data as outlined in an Institutional Review Board (“IRB”) approved protocol. Marketing campaigns for the Chromosome 8p Registry will be implemented as well as leveraging social media and partners clinicians, laboratories, and researchers to help with recruitment as approved by the IRB.

Gathering the Registry data is intended to facilitate research and help inform studies for potential experimental interventions. We anticipate research questions to include sponsored studies on treatment efficacy or how treatments work. The Registry data should lead to improved diagnostics and support care and meaningful clinical outcomes of a therapy.
Goal: Deliver a High-Quality Standard of Care

Focus Areas:

1. Build evidence to define high quality of care for 8p heroes

2. Manage and maintain a scalable Neurogenetics Multidisciplinary Clinical Research Network

3. Gather intelligence from families to inform patient needs and research priorities

Patients with chromosome 8p disorder need a roadmap to navigate all areas of diagnosis and prognosis, daily management, medical intervention, nutrition, resources, and cultivating a knowledgeable community. This is currently an urgent unmet medical need for our community.

Currently, they experience barriers to comprehensive care and disease-specific treatments. Challenges with delivery of high-quality care and therapeutic intervention include lack of access to a comprehensive team of experts, limited knowledge of disease courses, and a dearth of partnerships between disease stakeholders that requires coordination and commitment among all parties. Traditional health care involves local delivery of care. This model does not meet the needs of low density of patients living all over the world with a Chromosome 8p disorder. Consequently, diagnosis is delayed, and distance and disability limit access to comprehensive, ongoing care.

Affected individuals require multidisciplinary expert care. This is why we are funding a structured network of Neurogenetics Multidisciplinary Clinics nationally and ultimately internationally that will allow us to scale our efforts to achieve our vision. With a proper diagnosis, patients should be treated by a comprehensive team of collaborating specialists, including clinical specialists, genetic counselors, physical, occupational, and speech therapists, education advocates, and others based on patient and caregiver needs. This multidisciplinary approach will provide care in all main aspects of this disorder to include epilepsy, autism, developmental delays, muscle tone, and heart defects. With this approach, we will be able to collect information on the diverse aspects of this complex syndrome, with the purpose to further our understanding of this disorder and ultimately strive to develop treatment strategies. Clinics can help us integrate care and research, make sense of accessible electronic medical records, and identify objective outcome measures for meaningful
therapies for the patients. If we can characterize 8p disorders towards a standard of care, we can possibly prevent later onset of disease we may not yet know.

One way to ameliorate this is to leverage technology through teleresearch. Teleresearch, by its very nature, eliminates socioeconomic and geographic barriers to expert care, enables comprehensive data collection, improved understanding of natural history, and fosters improved patient–provider–researcher collaborations. In many ways, telehealth (i.e. the provision of health care remotely) is ideally suited for individuals with rare conditions addressing challenges of geography, travel burden, and access to experts. Telehealth allows us to expand the number of expert clinicians and providers globally to contribute to informed prognosis and care and educate caregivers on what to expect and how to manage them. We will also assess whether a dedicated chromosome 8p ICD diagnostic code would provide increased value for evidence based decision making and correlations in data tracking against other diseases.

Our strategy is driven by fostering a patient–clinician partnership that reflects the contributions of the patient as a disease expert.

Goal: Partner with Global Difference Makers

Focus Areas:

1. Solidify diversified and sustainable funding sources
2. Educate our investors about their value in our collective global impact
3. Create community activation partnerships

“Orphan drugs,” which are drugs that treat a rare disease, account for only 7.9% of money spent on developing and manufacturing drugs but increasing with the advancements in genomic precision medicine despite low demand.

Lack of funding and access to specialized clinics leads to a diagnostic odyssey -- consisting of repeated misdiagnosis, and delayed accurate diagnosis -- by doctors because of difficulty in assessing symptoms, access to genetic testing and counseling, and unfamiliarity with the wide spectrum of rare disorders.
We invite scientists, medical experts, rare disease organizations, and industry to collaborate with us to increase our chances for healthy, happy, and productive lives. By maximizing resources, funding, and best practices with each other, we can foster economies of scale which is the building block of positive change.

However, in the absence of an aggressive fund development plan, accessing care and subsequent treatment is very difficult, especially if seeking a multidisciplinary care team. Since rare diseases affect relatively few people, participation in studies and trials of different treatments is often limited. Limited research results in limited resources, which in turn impairs the clinicians’ ability to develop treatment plans for diagnosed patients.

We are inviting supporters to join us in being a Global Difference Maker to have an impact that leaves a legacy in science and medicine. This is an opportunity to be on the cutting edge of advanced genomics where there is a Global Commission to end the Diagnostic Odyssey so that every person with a genetic cause for an illness will have a diagnosis, allowing us to focus on treatment. We will continue to enlighten our donors as to how their donation makes an impact, inspiring them to partner with us in accomplishing this mission and demonstrating for them what their donations accomplish.

The four distinct groups that 8p will partner with for funding will include:

1. **Individuals**: The power of a peer-to-peer fundraising campaign is generated from our community and the Patient Leadership Board. A social network of friends, co-workers, and supporters can be exponentially powerful beyond dollars for Project 8p, but can also generate greater awareness and intimacy with our mission.

2. **Businesses/Corporations**: Our strategy to collaborate with corporations will require us to not only educate them about our mission but to also learn and understand their needs. We want to be aligned with organizations that share our values, goals, and ultimate vision. Companies that are building better businesses by valuing relationships with people, communities, and society to drive positive change. The incoming class of graduates entering the workforce as employees, consumers, and investors are taking note of good corporate actors and reward them with loyalty. Strong ESG values tend to boost employee motivation and attract talent through greater social credibility.

3. **Foundations**: Foundation partnerships is a critical area of support for us financially as well as in line with the grantor's mission. Moreover strategic foundations serve as partners in running support programs, providing complementary services to our community, and conduct direct charitable activities. Identifying funding mechanisms that align with Project 8p’s areas of focus and priorities will be the first step to applying and ultimately securing
grant awards. Beyond additional funding, we will have the chance to network and partner with the foundation further extending our reach.

4. **Government**: Federal, state and local government funding can meet many of our goals. As with grant funding, we will want to prioritize mechanisms that align with our initiatives. We could seek funding that bridges our goals with that of the broader community in basic and social sciences to translational and clinical research and infrastructure.

**Goal: Empower our Community**

**Focus Areas:**

1. Provide education and advocacy tools to the patient community

2. Significantly increase and maintain engagement by designing the user experience (“UX”) that transforms into a sustainable patient-led and patient-centered platform

3. Define content strategies for the patient community that connect to and influence the broader community

We are unique because of our patient led and patient centered approach. It unites our community of champions that have the power to be fierce advocates for 8p heroes and end the isolation of individuals coping with daily challenges. There are no greater experts than the families themselves living with the condition. With new programs to empower patients and families to be a partner in research priorities, advocacy, and services, Project 8p can help serve the needs of what matters most. Moreover, the organization’s Patient Leadership Board will provide clarity of the goals and serve as the voice of 8p heroes and caregivers through constant communication and education programs. For the external community, Project 8p will echo this voice of our 8p heroes and their families. We hope the inertia of self-motivation by individuals will move us closer to our goals together. This in turn will not only raise public awareness, but we can also work with decision-makers as partners enabling ownership of priorities determined by patients.
Project 8p believes scientists and clinicians, through patient engagement, have the obligation to gain a deeper understanding of how diseases impact the overall quality of life of patients and their families. Without the integration of a patient-centered process, we cannot deliver optimal quality care and treatment. We also want to reciprocally provide support back to patients and families to drive engagement. One form is providing data driven real-time insights back to families, therapists, clinicians, and researchers, and industry partners to identify treatments.

We want to help bridge the gap of socioeconomic and geographic barriers by facilitating structured exchange of experiences and ideas that can be an invaluable resource for rare disease families. We intend on designing a protocol to reach the international community with interactive structured webinar series with measurable outcomes. We will try to incorporate pragmatic therapies such as the following:

1. Special education
2. Physical therapy
3. Occupational therapy
4. Speech therapy
5. Feeding therapy
6. Behavioral therapy
7. Mobility aids
8. Alternative and Augmentative communication
9. Nutritional support

We can create Informational guides and expert discussions and access to augmentative and alternative communication devices and software can be transformative. A few examples of our successes are live streams and structured posts and virtual meetings with our Facebook Group, in person and hybrid conferences and gatherings, our Share What Works program, and informational webinars.

To facilitate a connection to the community, we will have a clearly defined content strategy with a primary focus on developing and maintaining relationships with our community of supporters. For the external community, Project 8’s engagement strategy will also focus on storytelling about our heroes, their families, and caregivers. We will strive to exchange information in a way that creates meaningful engagement by enabling our audiences to connect with those affected by 8p on a personal level.

We have started with understanding the everyday impact of the disease from those affected, and what they value most in terms of alleviation and expected outcomes. It is important to invest in a careful description of the clinical manifestations, disease course, and clinical outcomes. A comprehensive understanding of patient disease burden is also key to support the assessment and selection of the outcome measures that would provide a more meaningful life the 8p heroes deserve.
We are creating processes, organizational infrastructure, and events to support patients and
families to work in close collaboration with scientists, researchers, academia, and NIH. Project
8p will continue to provide the most up-to-date tools and resources to our community that
were previously unavailable.

_July 2021, Moving Mountains Conference, Westminster, Colorado_
Exhibit: Letter from the Founder

Dear Friends,

My daughter beams with light and gives hugs to everyone around her, from a stranger to her brother, so long as she senses positivity. Even all of her specialists (over a dozen!) receive a huge embrace with a cheek to cheek-smile from her. She is smart, and she captivates those around her.

She was born with what appeared to be healthy, except for a rare genetic mutation in the 8th chromosome. Some of her genes are missing, some are duplicated, and some are flipped around. And nobody has a clue as to how this happened.

When we first received the Chromosome 8p diagnosis, we were told, “Good luck, she’s not going to Harvard. You should seek parent counseling.” As you can imagine, there were endless tears that emptied the tissue box on the car ride home. And after, The 1st brochure we received from a genetics counselor came with the caveat that it was 30 years old. This diagnosis was so rare that they couldn’t tell me what her symptoms would be, all they could do was offer a range. The range was so broad, it spanned from minor global delays to wheelchair bound and questionable health. I was utterly scared and confused. All I kept thinking was, “That’s all you can tell me?”

That’s when I learned there was no central database for Chromosome 8p disorders or genetic conditions in this country, never mind the world. Months went by after the diagnosis with no info. Then I started scouring the internet. I found a couple of organizations and some basic databases. I started contacting any families I could find that were impacted by Chromosome 8.

Along the way, we decided to see our daughter for who she is and not what a brochure told us she could be. I have learned to forget about milestones and value the inch stones at her own pace, not mine.

When I hear my child’s voice with a faint “Hhhhi,” a dozen things go through my mind: my heart melts, I wonder how hard that must have been, I imagine neurons in her brain trying to send a message to the facial muscles, misfiring, and eventually her tongue moving and her breath producing that “hyyyyiii.” I am aware that it took 2 minutes for that motor plan, I am upset that it took that long, I am proud of her, I have tears of happiness and sadness. I can’t imagine how frustrating it must be when she has so many ideas to express and cannot, she wants to run alongside friends in the playground and cannot, and she wants to shout that she’s hungry and exhausted after a full day...and cannot.
But she can say hi, even if it took around 5 years and approximately 8,000 hours of all kinds of therapies. Could our journey to “hhhii” help other families? Could all of our collective experiences help each other if we only had a central repository of knowledge?

I am motivated to make sure my daughter can do everything she wants to do. That means coming together to find treatment. To help prevent others from being born with a genetic condition like this. To provide resources and a hand to hold for new families so that nobody needs to start from scratch and navigate the unknown like we did. To find a way to help the 8p heroes continue persevering with their beautiful smiles.

With Project 8p, I want to represent my daughter’s voice and its truth. I am her spokesperson, and one day, she will be the spokesperson for this foundation. I carefully listen to her communication cues, often it’s a wide eyed stare that sees right through my soul. I have full conviction that she is my teacher and has given me a purpose that is beyond our little family. As I say this out loud to her, she claps and reaches her arms out to embrace me.

So you see, I almost need her more than she will ever need me.

I hope when you read this, you can relate somehow. She is my inspiration to find a better, smarter way to respectfully fulfill her dreams.

And there are infinite possibilities.

With gratitude,

Bina Maniar Shah
Mother, Founder Project 8p