

Parental Perspectives on Living With a Child With HoPE

ELAINE E. STASHINKO,* LESLIE A. HARLEY, ROXANNE A. STEELE, AND NANCY J. CLEGG

This article describes the experiences and perceived needs of a small cohort of parents of children with holoprosencephaly (HPE). The factors that are important to the lives of children vary across families and stages of development. As children living with HPE grow and change, parents adapt their goals and expectations to reflect their child's now and future state. Relevant literature is integrated within the discussion to support recommendations for care. © 2010 Wiley-Liss, Inc.

KEY WORDS: holoprosencephaly; parents perception

How to cite this article: Stashinko EE, Harley LA, Steele RA, Clegg NJ. 2010. Parental perspectives on living with a child with HoPE. *Am J Med Genet Part C Semin Med Genet* 154C:197–201.

A medical diagnosis gives us insight into what a child has, but it is the child who truly shows us who he is.

Leslie Harley, President, Families for HoPE, Inc. and mom to Sammy who lived with semilobar HPE

While addressing the audience at the 1st NIH Conference on Holoprosencephaly (HPE), Dr. Francis Collins, Director of the National Human Genome Research Institute, noted that HPE is one letter short of spelling "hope." He challenged his audience to be the "O" team and find the missing letter needed to change HPE into HoPE. This article describes the experiences and perceived needs of a

While addressing the audience at the 1st NIH Conference on Holoprosencephaly (HPE), Dr. Francis Collins, Director of the National Human Genome Research Institute, noted that HPE is one letter short of spelling "hope." He challenged his audience to be the "O" team and find the missing letter needed to change HPE into HoPE.

small cohort of parents of children with HPE. Relevant literature is integrated within the discussion to support recommendations for care.

COMMON PARENTAL FEELINGS

Prenatal Diagnosis and Early Decisions

The cold and cloudy winter day of Friday, January 10, 2003, may not stand out in anyone else's history, but for me, it was the day that would change and define my life. It was the day I first heard the word holoprosencephaly.

Holoprosencephaly. 17 letters and 7 syllables. ho-lo-PROS-en-cef-a-le. We call it HPE for short.

We learned of Sammy's diagnosis during a routine ultrasound at 24 weeks gestation. We were told that he would most likely be still-born, and if he did survive to birth, he might not live long. We were told to expect severe mental retardation and facial deformities. We were told to be prepared to love a child who might never be able to show us his love in return.

Leslie Harley, President, Families for HoPE, Inc. and mom to Sammy

Elaine E. Stashinko is the Director of the Carter Center for Brain Research in Holoprosencephaly and Related Brain Malformations, Departments of Neurology and Developmental Medicine and Nursing, Kennedy Krieger Institute, Baltimore, MD.

Leslie A. Harley is the President of Families for HoPE, Inc. and mother to Sammy who lived with HPE for 4 years.

Roxanne A. Steele is a Member of the Board of Directors of Families for HoPE, Inc. and mother to Rachel, a 7-year-old living with HPE.

Nancy J. Clegg is the National Project Director of the Carter Centers for Brain Research in Holoprosencephaly and Related Brain Malformations, Department of Pediatric Neurology and Neurophysiology, Texas Scottish Rite Hospital for Children, Dallas, TX.

Grant sponsor: Carter Centers for Brain Research in Holoprosencephaly and Related Malformations; Grant sponsor: Don and Linda Carter Foundation; Grant sponsor: Crowley-Carter Foundation.

*Correspondence to: Elaine E. Stashinko, Ph.D., R.N., Department of Neurology and Developmental Medicine, Kennedy Krieger Institute, 801 North Broadway, Rm. 582, Baltimore, MD 21205. E-mail: stashinko@kennedykrieger.org

DOI 10.1002/ajmg.c.30237

Published online 26 January 2010 in Wiley InterScience (www.interscience.wiley.com)

Upon diagnosis, families are often told that their child has no chance for survival or meaningful interaction [Stashinko et al., 2004]. The first reaction of parents to a diagnosis of HPE, either prenatally or postnatally, is frequently a subjective feeling that the diagnosis is untrue. In a study by Redlinger-Grosse et al. [2002], factors influencing the decision to continue an affected pregnancy included the parents' religious and personal beliefs, past experiences, gestational age at diagnosis, and the uncertainty involved in the diagnosis of HPE. Some parents described feelings of isolation or lack of support from their providers, families, and friends because of their decision to continue an "abnormal" pregnancy.

One statement that I find myself asking other frightened expectant parents is, "Can you love him?" or "Can you love her?" When our fears take over and we don't feel we're equipped to care for a child with such challenges, we need to ask ourselves the basic question, "But, can I love him?" It is through that love that we do all of the things that we believe are impossible or unbearable.

Leslie Harley, President, Families for HoPE, Inc. and mom to Sammy

As disbelief subsides and parents begin to understand what it means to have a child with developmental disability, feelings of sorrow and fear as well as joy and hope may emerge. In a phenomenological study of the experiences of six parents of children with significant developmental disability, much of the parents' sorrow derived from their dealing with other people's negativity and hopelessness [Kearney and Griffin, 2001]. Parents came to know and love their child as they "are," and to find happiness in the little things that the child can do. Defiance of the prognoses that their children would "do nothing" enabled the parents to maintain hope and carryout therapeutic regimes. Similarly, living with a child with HPE requires balancing hopes and realistic expectations.

Living with a child with HPE requires balancing hopes and realistic expectations.

As one mother reflected:

When you have a child born with disabilities, first you grieve, then there is a dampening down and you scale down your expectations. You become more satisfied with less . . . we've learned to accept who he is and live with that on a day to day basis.

Vicki, mom to Michael, now 23-year-old young man with semilobar HPE

Parents are concerned about what may have caused the illness, and sometimes blame each other [Austin, 1990]. Parents must not only learn to cope with the illness and help the child cope, but often must also help the child's brothers, sisters, and grandparents cope—not easy tasks. Some parents cope by actively seeking information about this brain malformation, its symptoms, treatment, and prognosis.

When it came to Sammy's life, we often said that we were preparing ourselves for the worst but hoping for the best. We could quote the medical statistics of HPE backward and forward, so we were never in denial about the severity of the situation. We're sure that other people probably felt sorry for us, but we never felt sorry for ourselves. We made the best of it, and in return, we had the best time!

Leslie Harley, President, Families for HoPE, Inc. and mom to Sammy

A recent Swedish study of stress and well-being among parents of children with rare diseases found high parental stress, physical and emotional strain among mothers, especially single mothers [Delive et al., 2006]. Fathers demonstrated high stress related to incompetence. Both fathers' and working mothers' perceived knowledge and coping increased and stress decreased after an intervention aimed at empowering parents to manage their child's disability.

First being told that our daughter would not live more than a few months was devastating to us and going to check on her in her crib was terrifying because we were afraid to find her gone. Over time, as we learned she was doing well, we were able to turn that terror into triumph as physicians told us how well she was doing and we were doing as parents.

Roxanne, mother of Rachel, 7-year-old with semilobar HPE

As parents' knowledge about HPE and comfort in dealing with its symptoms grow, parents learn to accommodate their family activities to their child's physical condition and begin to develop strategies to handle problems as they arise. Some parents speak about having become stronger as a family unit, by supporting each other more and by realizing their strengths as a family.

Family adjustment to any chronic illness is a process. Parents are individuals also, and every family is unique. It is important to remember that each family member reaches acceptance at his or her own speed and in their own way. In this journey, every parent experiences emotional peaks and valleys. Feelings of sadness may surge around times of transition or expected developmental milestones such as when the child would have started walking or started school, or when younger brothers or sisters surpass their sibling [Shepard and Mahon, 1996].

Defining Quality of Life

Children with HPE experience symptoms that are variable, complex, and multifaceted. Increasingly, quality of life (QOL) is valued as an important addition to standard biomedical reports and as a measure of clinical progress and research outcome. Yet assessing QOL in families of children with HPE and other severe neurodevelopmental disabilities is challenging. The concept of QOL encompasses a broad range of domains including health and physical symptoms, social and psychological functioning, access to community resources, and satisfaction with medical care.

QOL for dependent children with or without disability necessarily includes the quality of caregiver care, acceptance, and adaptation. The quality of a child's life is interconnected with the quality of the environment in which they live. Children with disabilities live in a variety of housing, some with family and others in group homes or large institutional facilities. Their financial and community resources vary and differentially affect access to health and social services, educational and recreational programs [University of Toronto, 2003].

As part of a larger qualitative study of dimensions of QOL in children with neurodevelopmental disabilities, six mothers of children with HPE were interviewed (ES). All interviewed parents were asked: "When you think about defining quality of life for your child, what is important to you?" One mother of a 25-month-old child with semilobar HPE responded, "Parents determine goals and expectations. Our goal is Evan's ability to give and receive love, anything above that is a bonus."

When physicians speak about quality of life, I often want to ask them about their child's quality of life. Do they make it home every night before their child's bedtime? . . . My son with HPE knew his parents' love and affection because we lavished it upon him. Leslie Harley, President, Families for HoPE, Inc. and mom to Sammy

A recurring response was the importance of the child's ability to socially interact and participate in the world around him. One parent stated that her daughter's "ability to respond to touch" and "recognize family and caregivers" was an important indicator of well-being (Liz, mom to 6 year old with alobar HPE).

He will never be able to tell me he loves me in the way that typical children can. He can simply look at me, and we can speak to each other with our eyes and our hearts. He is my son. I am reminded on a daily basis that he is severely brain damaged, and I have faced that reality head on. As a mother, I feel

helpless at times that I cannot advocate strongly or loudly enough for my child. As a parent, do you know how touching it is to seek help and have so many people at least attempt to help you? To have people of power attempt to go to the end of the Earth for your child? As a parent of a severely disabled child, I must remind myself day after day and hour after hour that "love never fails."

Leslie Harley, President, Families for HoPE, Inc. and mom to Sammy

How each of us defines QOL varies among families and within families at different points of time. QOL for all people reflects how satisfied they are with aspects of life that are important to them [University of Toronto, 2003, p. 8; Ruta et al., 1994]. All the parents described social acceptance—feeling their child was accepted within the family and medical community and treated respectfully by others—as important.

DISCUSSION AND RECOMMENDATIONS

Combat Myths and Misconceptions With Knowledge and Hope

Misinformation about HPE and its treatment and prognosis persists among healthcare providers. While there is a wide spectrum of abilities and experiences in families of children with HPE, much literature still categorizes HPE as a fatal diagnosis. Many parents of children with HPE have been told to expect their child to die within a few days or months after birth, and then need to readjust their expectations to live with a surviving child and his/her clinical outcomes.

While there is a wide spectrum of abilities and experiences in families of children with HPE, much literature still

categorizes HPE as a fatal diagnosis. Many parents of children with HPE have been told to expect their child to die within a few days or months after birth, and then need to readjust their expectations to live with a surviving child and his/her clinical outcomes.

The Carter Centers for Brain Research in Holoprosencephaly (<http://www.stanford.edu/group/hpe/>) have enrolled 182 children in a 10-year longitudinal International Database study; children enrolled in this database have HPE confirmed by brain scan review. The mean age of the children with HPE in this database is 9.4 years, with a range from 3 months to 28 years of age. The mean age of the 27 children with the most severe form of HPE, alobar HPE, is 5.9 years (range 3 months to 20 years). In our experience, most children with classic HPE develop minimal motor and language skills, and most children with MIH (the least severe HPE category) walk with assistance, speak and function with mild cognitive impairment [Lewis et al., 2002; Plawner et al., 2002]. Therefore, when a fetus or child is diagnosed with HPE, it is important that pediatricians and obstetricians who help families make decisions involving pregnancy care, interruption and/or birth provide accurate information about the spectrum of clinical outcomes [Levey et al., 2010; Stashinko et al., 2004].

In a Mayo Clinic article, Li [2000] stated that hope is essential to the healing and recovering process. Sensitive honest communication, which maintains hope, is crucial in promoting parental strength and coping [Kearney and Griffin, 2001]. In rare disorders like HPE where science has much to learn, hope enables families to forge new frontiers and create new possibilities.

Sensitive honest communication, which maintains hope, is crucial in promoting parental strength and coping [Kearney and Griffin, 2001]. In rare disorders like HPE where science has much to learn, hope enables families to forge new frontiers and create new possibilities.

Networking

Sharing fears and concerns with other parents of children with HPE, for example, on the HPE listserv (<http://groups.yahoo.com/group/holoprosencephaly/>), helps parents know that they are not different or alone. Networking with other families is also a great way to share resources and solutions to everyday problems and common challenges. “Veteran” parents who have learned to effectively negotiate the system may provide immense support and role modeling to “novice” parents of children with HPE. Parent support groups are particularly helpful near the time of diagnosis, probably because of the need for concrete information that is shared among a relatively small number of people who are in a similar situation [Shepard and Mahon, 1996]. The benefits of parent networking are what led a small group of mothers to formalize a support organization called Families for HoPE, Inc. (<http://familiesforhope.org/>).

Families for HoPE works to help families find understanding and hope in a world full of negatives. Being able to talk to other parents, receive on-going support, and share each child's story helps families to come to terms with the HPE diagnosis and look to quality of care and life.

Roxanne, Families for HoPE Board and mom to Rachel

Identify Sources of Support

Health professionals can assist families identify sources of emotional support and practical help. Findings of a recent study on parental coping underscore the importance of helping parents of children with physical disabilities maintain social support through community resources [Cavallo et al., 2009].

It truly took a village at times to raise him, but Sammy didn't just take from society; he contributed to the betterment of society. He was involved in the community—raising funds for his preschool, his church, his hospital, and his condition.

Leslie Harley, President, Families for HoPE, Inc. and mom to Sammy

Types of support that families have found helpful include transportation for siblings, having siblings stay with friends during hospitalizations, providing meals, doing the laundry, running errands, or baby-sitting in the home so that the parents can have some time for themselves [Shepard and Mahon, 1996].

Focus on the Child's Strengths and Positive Qualities

Each child is unique, with his or her own abilities and possibilities. Parents take pride in their children's survivorship and resilience.

In our experiences, our daughter is writing her own story and she does not conform to the medical books or past experiences of her physicians.

Brian, father of a 7-year-old with semilobar HPE.

Develop Realistic Goals

After a prenatal diagnosis, professionals can help parents negotiate their way through additional medical visits by helping them foresee the range of possible needs for the baby, including preparing for possible death or birth of a child with urgent medical needs [Redlinger-Grosse et al., 2002].

Postnatally, providers and parents need to partner to identify meaningful, attainable goals. Management of symptoms, such as altered sleep cycles, can be just as important to families as seizure management. Encouraging parents to think about and verbalize their care priorities can serve as a starting point for a parent-clinician dialog about treatment goals and future care. As with any child, many goals will be reached and many will not. However, every child will progress.

CONCLUSION

The factors that are important to the lives of children vary across families and stages of development. As children living with HPE grow and change, parents adapt their goals and expectations to reflect their child's now and future state. Every child is different and special, and every child writes their own story. Most of us have come to understand that meaning and QOL are not determined by physical function alone or the length of time one lives, but rather the quality of one's relationships. Forging parent-clinician partnerships with mutual respect and ongoing dialog allows us to work together toward hopeful futures.

Sammy was a fighter from the very start and a boy of unbelievable courage. He was a wonderful gift and blessing. He was a little boy who couldn't walk or talk, yet he touched hearts. His body was weak, yet his spirit was strong. He inspired others to love deeply and to give more of themselves, yet he never asked for anything in return.

On the night that my son passed away after a 5-year journey with HPE, two physicians came to the hospital to comfort us as we said goodbye to our son—his pediatrician and his neurologist. Before leaving for that final time, my mother took a moment to ask the neurologist what he had learned from my son. His response was simply, “Humility. I learned humility.”

Leslie Harley, President, Families for HoPE, Inc. and mom to Sammy

ACKNOWLEDGMENTS

Special thanks to Families for HoPE and all the parents who participated in this research and manuscript preparation. The research was supported by the Carter Centers for Brain Research in Holoprosencephaly and Related Malformations, the Don and Linda Carter Foundation, and the Crowley-Carter Foundation.

REFERENCES

- Austin J. 1990. Assessment of coping mechanisms used by parents and children with chronic illness. *MCN* 15:98–102.
- Cavallo S, Feldman DE, Swaine B, Meshefedjian G. 2009. Is parental coping associated with the level of function in children with physical disabilities? *Child Care Health Dev* 35:33–40.
- Delive L, Samuelsson L, Tallborn A, Fash A, Hallberg LR. 2006. Stress and well-being among parents of children with rare diseases: A prospective intervention study. *J Adv Nurs* 53:392–402.
- Kearney P, Griffin T. 2001. Between joy and sorrow: Being a parent of a child with developmental disability. *J Adv Nurs* 34: 582–592.
- Levey EB, Stashinko E, Cleggy NJ, Delgado MR. Management of children with holoprosencephaly. *Am J Med Genet Part C Semin Med Genet* 154C:183–190.
- Lewis AJ, Simon EM, Barkovich AJ, Clegg NJ, Delgado MR, Levey EB, Hahn JS. 2002. Middle interhemispheric variant of holoprosencephaly: A distinct cliniconeuroradiologic subtype. *Neurology* 59:1860–1865.
- Li J. 2000. Hope and the medical encounter. *Mayo Clin Proc* 75:765–767.
- Plawner LL, Delgado MR, Miller VS, Levey EB, Kinsman SL, Barkovich AJ, Simon EM, Clegg NJ, Sweet VT, Stashinko EE, Hahn JS. 2002. Neuroanatomy of holoprosencephaly as predictor of function: Beyond the face predicting the brain. *Neurology* 59: 1058–1066.
- Redlinger-Grosse K, Bernhardt BA, Berg K, Muenke M, Biesecker BB. 2002. The decision to continue: The experiences and needs of parents who receive a prenatal diagnosis of holoprosencephaly. *Am J Med Genet* 112:369–378.
- Ruta DA, Garratt AM, Leng M, Russell IT, MacDonald LM. 1994. A new approach to the measurement of quality of life: The Patient-Generated Index. *Medical Care* 32:1109–1126.
- Shepard M, Mahon M. 1996. Chronic conditions and the family. In: Jackson PL, Vessey J, editors. *Primary care of the child with a chronic condition*. 2nd edition. St. Louis, MO: Mosby-Year Book, Inc. pp 41–57.
- Stashinko EE, Clegg NJ, Kammann HA, Sweet VT, Delgado MR, Hahn JS, Levey EB. 2004. A retrospective survey of perinatal risk factors of 104 living children with holoprosencephaly. *Am J Med Genet Part A* 128A: 114–119.
- University of Toronto. 2003. Quality of Life Model. The Quality of Life Research Unit, Centre for Health Promotion, Department of Public Health Sciences. Retrieved from: <http://www.utoronto.ca/qol/>.