WOLF-HIRSCHHORN SYNDROME [WHS]

PRIMARY RESEARCH FOCUSES

Developmental Disability / Feeding Difficulties / Seizures

John Carey, MD, Professor of Pediatrics , University of Utah

WHS is caused by distal deletion of the short arm of the 4th chromosome. Often times, WHS is loosely referred to as 4P- (4th chromosome, petit arm, missing).

Frequency of WHS is estimated to be 1 in 50,000 live births or a little more than 1 birth per week in the United States.

Only 20% of the cases are a result of one parent being a carrier. The remaining cases are rare chance occurrences.

Phenotype (characteristics) of WHS include:

- Slow Growth
- Developmental delays
- Cranial/facial distinctions
- Closure defects including cleft lip or palate
- Cardiac defects
- Low muscle tone
- Kidney defects
- Microcephaly
- Epilepsy

Abilities range from those who are in need of constant care to those who are verbal, mobile, and interactive.

For additional WHS clinical information visit:

http://www.ncbi.nlm.nih.gov/books/NBK1183

SHARING EXPERIENCES

The 4P- Support Group offers a variety of opportunities for families to obtain information and support in the care of their child with WHS.

National gatherings take place biannually. This is an opportunity for families and caregivers to come together to attend workshops, network with other families, and celebrate their special children.

Regional gatherings take place opposite the national gathering. This allows networking and sharing of therapeutic tips and tricks.

An email listserv is available for parents, families and interested professions. This allows opportunities for questions, answers, concerns and experiences to be shared.

Newsletters are distributed quarterly to members of our support group.

Biographies are available from the support group. A nominal fee may apply.

Support groups are active in North America, United Kingdom, Australia, Italy, Germany, & France.

www.4p-supportgroup.org

BUILDING FUTURES

The support group has more than 600 registered families. Medical literature cites a fraction of these numbers.

Together with Dr. John Carey (U.S. renowned WHS expert) and international geneticists, we hope to bring more awareness of this syndrome to others. Future national gatherings are planned to coincide with medical genetics conferences.

WHS families have participated in an array of research studies around the world. However, further medical and therapeutic research is needed.

Our goal is to optimize the information needed to provide the best care for our children.

Improvements in genetic testing will result in an increase in the number of WHS diagnoses and genetic chip technology advances will reveal greater detail of the human genome. Both advances will provide increased research opportunities.

The support group strongly encourages medical professions to contact the group concerning research participation.