

Care Recommendations for individuals with 4p-

Dr. Amy Calhoun, Medical Geneticist Medical Advisor to the Board of Directors of the 4p- Support Group

To do in early life/soon after diagnosis/at the time of initial evaluation if possible:

- Electroencephalogram (EEG), Ideally a 72-hour video EEG
- Echocardiogram and EKG
- GI: Feeding, reflux, aspiration, slow motility
- Detailed eye exam
- Hearing check, ideally with a brainstem auditory evoked response
- Immune system screening: immunoglobulin levels, lymphocyte subsets, polysaccharide responsiveness
- Complete blood count (CBC)
- Kidney evaluation: BUN, creatinine, cystatin-c, urinalysis, renal ultrasound
- Skeletal evaluation
- Developmental evaluation
- Sleep study: Sleep apnea is common in WHS

Ongoing care:

- Regular neurology follow-up with EEGs as needed
- Annual CBC
- Annual kidney testing: BUN, creatinine, cystatin C, urinalysis
- Ongoing comprehensive developmental and rehabilitation support: communication, speech, feeding, PT, OT, school, adult transition preparation
- Routine skeletal exams for scoliosis especially in adolescence
- Other subspecialty evaluations as needed: GI, cardiology, pulmonology, immunology, endocrinology, etc.

May consider these additional evaluations:

- Liver ultrasound at diagnosis and yearly thereafter
- Baseline and yearly kidney ultrasound
- Growth hormone stimulation test

For additional management information visit:
<https://www.ncbi.nlm.nih.gov/books/NBK1183>

Also visit www.4p-SupportGroup.org.