



VHLA Suggested Active Surveillance Guidelines

Until a cure is found, surveillance is a patient's strongest defense to prevent severe VHL complications.

[For references, click [here](#). Revised 5/20/16]

Surveillance is the testing of individuals at risk for von Hippel-Lindau disease (VHL) who do not yet have symptoms, or who are known to have VHL but do not yet have symptoms in a particular area. The unaffected organs should still be screened.

Modifications of surveillance schedules may sometimes be done by physicians familiar with individual patients and with their family history. Once a person has a known manifestation of VHL, or develops a symptom, the follow-up plan should be determined with the medical team. More frequent testing may be needed to track the growth of known lesions.

People who have had a DNA test and do not carry the altered VHL gene may be excused from testing. Even with the VHL gene, once an individual has reached the age of sixty and still has no evidence of VHL on these surveillance tests, imaging tests may be reduced to every two years for MRI.

Revisions in this version of the surveillance guidelines for VHL include a change in recommendations from CT to MRI, in order to reduce exposure to radiation for all people. CT should be avoided for all pre-symptomatic people, and should be reserved for occasions when it is truly needed to answer a diagnostic question.

In order to monitor the most critical areas of the brain and spinal cord in the most efficient and cost-effective manner, CNS MRIs should include the brain, cervical, thoracic, and lumbar spine. Scans should be ordered as no less than a 1.5T MRI with and without contrast, with thin cuts through the posterior fossa, and attention to inner ear/petrous temporal bone to rule out both ELST and hemangioblastomas of the neuraxis.

Regular audiometric tests are included in the surveillance protocol to provide a reference point in case of sign or symptom of hearing loss, tinnitus (ringing in the ears), and/or vertigo (dizziness, loss of balance). If hearing drops, swift action may be required to save hearing.

MRI is the preferred surveillance method for the abdomen. Quality ultrasound may be substituted for MRI of the abdomen no more than once every two years. "Quality" is defined as a machine that produces good quality pictures, with an operator experienced in imaging the organs being studied. The objective is to find even small tumors, which are difficult to identify on ultrasound.

Any Age

Inform families that, if they choose, they and their geneticist may contact one of the clinical DNA testing laboratories familiar with VHL for DNA testing. If the family marker is detectable, DNA testing can identify those family members who are not at risk and may discontinue surveillance. Testing may also be useful in calculating risks for family members who do carry the altered gene and require periodic surveillance tests. Risk factors are not definitive indicators of what will happen, but only highlight areas at higher or lower risk probability. Early detection and appropriate treatment are the best defenses.

From Conception

Inform obstetrician of family history of VHL. If the mother has VHL, see also the discussion of pregnancy in the VHL handbook and in the surveillance protocol. A woman who is having any genetic testing done may request a VHL test be included in that series of tests.

From Birth

Inform pediatrician of family history of VHL. Pediatrician to look for signs of neurological disturbance, nystagmus, strabismus, white pupil, and other signs which might indicate a referral to a retinal specialist. Obtain routine newborn hearing surveillance.

Ages 1-4

Annually

- Eye/retinal examination with indirect ophthalmoscope by an ophthalmologist skilled in diagnosis and management of retinal disease, especially for children known to carry the VHL mutation.
- Pediatrician to look for signs of neurological disturbance, nystagmus, strabismus, white pupil, and abnormalities in blood pressure, vision, or hearing.

Ages 5-15

Annually

- Physical examination and neurological assessment by pediatrician informed about VHL, with particular attention to blood pressure (taken while lying down and standing), hearing impairment, neurological disturbance, nystagmus, strabismus, white pupil, and other signs indicating retinal problems.
- Dilated eye/retinal examination with indirect ophthalmoscope by ophthalmologist informed about VHL.
- Test for fractionated metanephrines, especially normetanephrine in a “plasma free metanephrine” blood test or in a 24-hour urine test. Abdominal ultrasonography annually from 8 years or earlier if indicated. Abdominal MRI or MIBG scan only if biochemical abnormalities found.

Every 2-3 Years

- Audiology assessment by an audiologist. Annually if any hearing loss, tinnitus, or vertigo is found
- In the case of repeated ear infections, MRI with contrast of the internal auditory canal using thin slices, to check for a possible ELST.

Age 16+

Annually

- Physical examination by physician informed about VHL.
- Dilated eye/retinal examination with indirect ophthalmoscope by ophthalmologist informed about VHL.
- Quality ultrasound and at least every other year when not pregnant, an MRI scan) of abdomen with and without contrast to assess kidneys, pancreas, and adrenals.
- Test for fractionated metanephrines, especially normetanephrine in “plasma free metanephrines” blood test or 24-hour urine test. Abdominal MRI or MIBG scan if biochemical abnormalities found.

Every 2-3 Years

- MRI scans should be ordered as no less than a 1.5T MRI with and without contrast of brain, cervical, thoracic, and lumbar spine, with thin cuts through the posterior fossa, and attention to inner ear/petrous temporal bone to rule out both ELST and hemangioblastomas of the neuraxis.
- Audiology assessment by an audiologist.

During Pregnancy (for women with VHL)

- Regular retinal checkup to anticipate potentially more rapid progression of lesions.
- Test for pheo early, mid, and again late pregnancy to ensure no active pheo during pregnancy or especially labor and delivery.
- During the 4th month of pregnancy, MRI—without contrast—to check on any known lesions of the brain and spine. If known retinal, brain, or spinal lesions, consider C-section.

Updated 7/13/2017