



Evaluation Guidelines for an Individual with PMS and Ring 22 for NF2

Individuals with ring 22 are at increased risk of developing NF2 due to the instability of the ring chromosome and the risk of a subsequent mutation at the NF2 locus at 22q12.2 (the two-hit pathway). The ring chromosome is unstable during cell division and can be lost as the mitotic cells divide, resulting in daughter cells that contain only a single copy of chromosome 22 (first hit). If a somatic mutation occurs at the NF2 locus on the existing copy of chromosome 22 (second hit), the individual is at risk of NF2 and should be monitored for symptoms.

Surveillance:

- Cranial MRI should begin between ages 10 and 12 years and continue annually until at least the fourth decade of life. The age at which monitoring can be safely stopped is unclear.
- Consider spinal MRI every 3-5 years or as indicated by symptomatology
- Hearing evaluations, including BAER testing, may detect changes in auditory nerve function before changes can be visualized by MRI.
- Complete eye exams should be conducted annually.
- Cutaneous examination.

Evaluations following the diagnosis:

Once a diagnosis of NF2 is made, the extent of disease should be established by the following testing, if not already performed:

- Cranial MRI
- Hearing evaluation, including brain stem auditory evoked response (BAER)
- Ophthalmologic evaluation
- Cutaneous examination
- Consider spinal MRI

Please note: Evaluation and treatment of individuals with NF2 are best undertaken in an NF2 center experienced in managing the manifestations of this condition. Once the diagnosis of NF2 is established, consultation with neurosurgery and oncology may be warranted.

For more comprehensive information on the surveillance, management and treatment of individuals with NF2, please consult the following reference:

Neurofibromatosis 2, Gene Reviews – March, 2018 –
<https://www.ncbi.nlm.nih.gov/books/NBK1201/>

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