



MELORHEOSTOSIS AT NIH

National Institutes of Health (NIH) is conducting an observational natural history study of the progression and cause of Melorheostosis in adults with an emphasis on genetic, endocrine and clinical manifestations of the disease. This is a clinical study in which patients with melorheostosis are enrolled, receive routine clinically indicated tests, and are followed over time. Testing and evaluation are highly individualized based on a patient's age, symptoms and past evaluations. A comprehensive physical exam, medical history, health related quality of life questionnaires, laboratory and radiological evaluations will be done during a 3 to 4 day work up at the NIH. Additional follow-up visits may be required, and then followed by NIH visits every other year. Since melorheostosis presents slowly and symptoms can vary greatly depending on the extent and location of excess bone growth, the testing strategy will be tailored to the individual patient as medically indicated. Radiation exposure will be limited to medically indicated tests.

Purposes of the tests are:

1. To establish a firm diagnosis of Melorheostosis,
2. To determine the extent of the disease and any symptoms,
3. To evaluate any pain source such as impingement on nerves,
4. To exclude other diagnoses which cause excessive bone disease,
5. To guide treatment.

Laboratory and clinically indicated scans:

1. Blood tests for complete blood counts, chemistry, inflammation markers, bone turn-over markers, pregnancy test (in females of reproductive age prior to performing any radiological studies) bone metabolism and 24hr urine collection
2. Bone scan to assess metabolic activity of the lesions
3. Specific x-rays of involved bones
4. Magnetic Resonance Imaging (MRI) for better delineation of soft tissue lesions which may be impinging on nerves, if needed.

Consults as needed:

1. Orthopaedic evaluation.
2. Dermatology consult to establish a baseline skin exam and examine for skin lesions possibly associated with melorheostosis.
3. Neurological evaluation will be available on all patients.
4. Pain and Palliative Care to help qualify the nature of pain and identify potential therapy for pain management.
5. Physical and Rehabilitation Medicine may consult to provide patients a way to optimize their physical fitness, prevent secondary complications, and improve quality of life. All

patients will be evaluated by Physical therapy (PT), Occupational therapy (OT) and Recreational Therapy (RT).

6. Clinical photos may be obtained to record skin lesions or joint changes. The face and identifiable features will be blacked out.

7. Dermatology consult will be obtained. If there are skin changes over the affected bone, a skin biopsy will be done to test for the known genetic markers for bone disease.

Frequently Asked Questions

Who is eligible for the study?

All adults (18 and older) with a presumptive diagnosis of melorheostosis.

Where is the study being conducted?

At the NIH Clinical Center in Bethesda, MD. Located just outside Washington DC, the Clinical Center is the nation's research hospital. <http://www.cc.nih.gov/>

Are there any charges to patients? No. All costs for consultation and testing at NIH, as well as room, board, and parking, will be paid by NIH. There will be no charges to patients.

Is participation limited to patients in the United States? No, adult melorheostosis patients from around the world are eligible to apply.

Who pays for travel? Patients are responsible for any travel related cost to NIH. Should you choose to drive to NIH, free parking is conveniently available at the Clinical Research Center for all patients.

Will you coordinate with my doctor? Yes, we will provide a report to your physician (with your permission), and copies of your test results upon request. Results of the tests performed are also available on the internet through the Patient Portal for your access: <https://patientportal.cc.nih.gov>

Will the tests be repeated at each visit?

The testing will not be repeated at each visit. The testing regimen will be tailored to each patient, and only the minimum tests performed to look for new melorheostotic lesions or progression.

How can I get more information?

Visit Clinicaltrials.gov, or contact Nancy A. Spencer, RN/MSCN at nancy.spencer@nih.gov.