

Seeing the signs: Acromegaly

Acromegaly is a rare disorder typically caused by a growth hormone (GH)-producing tumor in a small gland at the base of the brain: the pituitary. This organ is a “master gland”, controlling the production and release of many different hormones throughout the body. In children, GH released from the pituitary is responsible for growth, and in adults it helps to regulate the metabolism of muscles, bones and lipids. In acromegaly, the body is flooded with an excess of this hormone and, over time, this causes many problems, both physical and physiological. The outward symptoms of acromegaly are distinctive: a person with the disorder will often develop coarse facial features, including a protruding brow and jaw, in addition to large hands and feet. Acromegaly usually also causes a number of health-related symptoms, such as joint pain, sleep apnea, carpal tunnel syndrome, and fatigue, as well as growth of the inner organs, e.g. the heart. These latter problems are particularly concerning, as they can be the cause of premature death in patients with acromegaly.

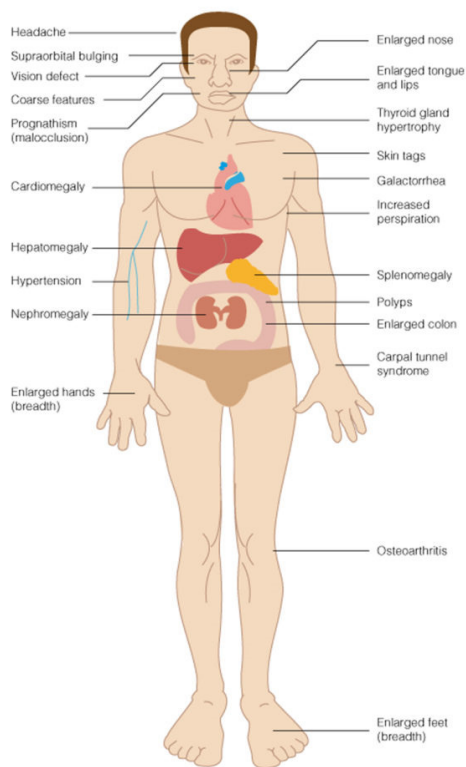


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Fig. 1. (Drafting of summary and illustration kindly supported by IPSEN, Germany.)

As the physical symptoms of acromegaly develop gradually, one of the most difficult issues for any patient is to have the correct diagnosis made: close relatives or the family physician may not notice the changes that progress insidiously over months and years. Each year, five people per million are

diagnosed with acromegaly worldwide. Many of them wait for years to receive the diagnosis “acromegaly” as a prerequisite for disease-directed treatment such as pituitary surgery. Although we do not yet have a clear idea of why this delay occurs, we do know that early diagnosis and treatment are crucial for alleviating symptoms, preventing the development of other disorders and early death.

To be able to overhaul the diagnostic process of acromegaly, we needed to know where the problem lies. We chose to look at the process from the patient’s perspective: we surveyed 165 patients with acromegaly about their experiences before diagnosis. Our most striking finding was that, on average, patients waited 2.9 years for a diagnosis after a first visit to the doctor with acromegaly-related symptoms, and visited 3.4 physicians before being diagnosed correctly. Patients visited different specialists for the treatment of symptoms that can occur in conjunction with acromegaly; for example, they saw the neurologist for carpal tunnel syndrome, the orthopedist for joint pain, and the ear, nose and throat specialist for snoring. However, by the isolated presentation of specific symptoms, the consulted physicians have little hope of correctly “connecting the dots”. Yet, without a holistic diagnosis, the patient is left with the treatment of an individual symptom, rather than a comprehensive care plan for the underlying illness. This, in turn, leads to “doctor shopping” in the hope that the next expert will provide a treatment that works. We found women to be more affected by this than men, as they typically waited longer for a correct diagnosis (4.1 years versus 1.5 years, respectively), and visited more doctors in the process (4 versus 2.7, respectively).

So, how do we prevent patients from seeking symptom-specific medical treatment, and doctors from providing symptom-specific care? Our study suggests one answer lies with the physicians – specifically by increasing their awareness of acromegaly symptoms and their interrelatedness. By having a more holistic understanding of the disorder, a doctor should more easily recognize typical symptom combinations. We believe by having a medical community who are better educated about acromegaly symptoms, we will see decreased diagnostic times and thus increased speed of treatment. We hope that, in turn, this will lead to better disease management and an eventual easing of the burden of this disease.

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Publication

[Diagnosis and management of acromegaly: the patient’s perspective.](#)

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