INTRODUCTION:

Multidisciplinary, the management of dwarfism relies on several medical professionals. This factsheet aims to shed light on the main specialists that a little person or members of their support system will encounter.

GENETICIST:

Medical genetics takes care of testing and the analysis of physiological and pathological conditions associated with genetic diagnoses, which concern the hereditary background of the individual. A geneticist is a specialist concerned with the investigation, diagnosis and management of people with inborn errors of metabolism, chromosomal abnormalities and dysmorphisms (irregularities). The geneticist thus manages various conditions including congenital malformations, sensory impairments (hearing loss, blindness, etc.), learning disorders and developmental delay, metabolic errors, growth problems (mild or severe) as well as family diseases (cancer, high cholesterol levels, heart disease, etc.). The geneticist is also able to provide genetic counseling and perform prenatal screening tests.

For little people, the geneticist is a key person in identifying the diagnosis causing the short stature. For parents with dwarfism wishing to have a child, it will guide their approach by offering them genetic counseling. This process provides information about the nature and possible outcomes of their decisions, the likelihood of passing their diagnosis on to their child, and the options available to them regarding life and family planning.

ORTHOPEDIST:

The orthopedist is a doctor authorized to medically and/or surgically treat pathologies of the musculoskeletal system. They treat diseases of the bones, joints, ligaments, muscles, tendons and nerves. The orthopedist is therefore a specialist in the treatment of fractures, dislocations, ligament injuries (sprains), herniated discs, bone tumors and other similar diagnoses. They also manage congenital malformations (club foot, congenital dislocation of the hip, etc.), growth anomalies, and the sequelae (longer term or chronic after effects) of infectious or inflammatory diseases. Some orthopedists specialize in hip or leg problems, and others with spine issues.

For little people, the orthopedist turns out to be an essential ally in treating various musculoskeletal problems related to dwarfism, such as clubfoot, scoliosis (lateral curvature of the spine), kyphosis (backward curvature), lordosis (forward oriented curvature of the spine),
straightening of varus or valgus (bowing of the legs), treatment of herniated discs (in concert with a neurosurgeon) or even replacement of joints (hips and knees in particular). In addition, the orthopedic surgeon will be able to perform limb lengthening, a controversial treatment, for those who wish to consider such a procedure.

**PHYSIATRIST:**

The physiatrist is a doctor who specializes in the diagnosis, treatment and rehabilitation of conditions of the musculoskeletal system. Close to rheumatology and orthopedics, physiatry is distinguished by its diagnostic methods which place great emphasis on clinical examination. Its therapeutic methods rely on manual intervention, physical agents, pharmacological agents and various types of management. Physiatry also places great emphasis on the functional impact of a pathological condition and its rehabilitation, often using physiotherapy and occupational therapy. The main disorders treated in physiatry offices are back and neck pain, rheumatic disorder, arthritis and other joint problems, sequelae (longer term or chronic after effects) of fractures or sprains, neuritis and neuralgia, muscle disorders, and certain headaches, etc. In a hospital setting, the physiatrist takes care of all diagnoses of the musculoskeletal system in the arena of rehabilitation.

Therefore, for little people, the physiatrist will in particular be called upon to relieve back and cervical disorders. They may, for example, offer interventions to treat herniated discs (if these do not require surgery). It will also be useful in managing joint problems in the hip or knee. The physiatrist will occupy a prominent place in rehabilitation efforts.

**NEUROSURGEON:**

Neurosurgery is a surgical specialty that manages disorders of the central, peripheral and autonomic nervous system. The neurosurgeon is involved in the treatment of patients struggling with disorders of the nervous system (diagnoses of the brain, meninges, cranium and its blood supply), conditions of the pituitary gland, issues with the spinal cord and of the spine, including conditions requiring treatment by spinal fusion or instrumentation.

Like the orthopedist, the neurosurgeon will be present to help manage medical issues for people of short stature when they have to undergo operations related to complications from their diagnosis. For example, the neurosurgeon may treat macrocrania (abnormally large skull), hydrocephalus (accumulation of cerebrospinal fluid in the brain), bulbocervical compressions, craniocervical instability, spinal stenosis (narrowing of the spinal canal) and herniated discs in consultation with an orthopedist.
OTOLARYNGOLOGIST:

The otolaryngologist, cervico-facial surgeon, specializes in the treatment of head and neck problems. They perform a variety of surgeries of the ear, nose, sinuses, pharynx, larynx, oral cavity, neck, thyroid, salivary glands, bronchi and esophagus. Trained for ear surgery, they can restore hearing through microsurgery. This specialist also employs other surgical techniques to cure or treat illnesses, infections or birth defects. In addition, their training in cervico-facial surgery allows them to perform cosmetic surgery. This reconstructive technique is applied to the nose, ears, jaw and face to improve or restore function and appearance.

Regarding little people, the otolaryngologist will mainly treat recurrent ear infections and problems with hearing loss or deafness. They will also take care of language and communication issues that may arise. In addition, this specialist may perform an adenoidectomy (removal of adenoids) and/or a tonsillectomy (removal of the tonsils) to treat obstructive sleep apnea.

PSYCHOLOGIST:

The psychologist is an expert in behavior, emotions and mental health. They work with people who experience distress or psychological symptoms. The main task of the psychologist is to offer psychotherapy to their patient to help them feel better, find answers to their questions, solve their problems, make choices and understand themselves better. Psychotherapy is considered a treatment for a mental diagnosis, for behavioral disturbances or for any other problem causing psychological distress or suffering. Its objective is to promote within the person significant cognitive improvements, better emotional or behavioral functioning in their interpersonal relationships, and to help with their personality or state of mental health. This treatment goes beyond helping to cope with common difficulties or providing counseling or support.

The psychologist can be a great support for little people and those around them. With parents of average height who have given birth to a child with small stature, psychotherapy can help them to adapt psychologically to caring for a child with the diagnosis and come to terms with it, and to cope with their feelings. Regarding little people, psychotherapy can help them accept their condition and their body, improve their self-esteem and self-confidence, overcome feelings of shyness or improve their interpersonal relationships (place in the family, romantic relationships, etc.). It is also effective in treating depression, anxiety, and in learning to manage chronic pain.
OTHER SPECIALISTS:

- **Endocrinologist:**
  Endocrinology is the science that studies glands and hormones. Hormones play a central role in several basic functions such as growth, development, reproduction and energy production. The endocrinologist will be particularly useful for little people with pituitary conditions (such as pituitary insufficiency), diagnoses related to growth hormones or those related to bone density.

- **Pulmonologist:**
  Pulmonology is the medical specialty that deals with respiratory problems (diseases of the lungs, bronchi and pleura). Respiratory therapy also treats symptoms such as chronic cough, sleep apnea, shortness of breath, or allergies. Thus, little people who suffer or are at risk of developing sleep apnea, obstructive apnea or restrictive respiratory syndrome should consult a pulmonologist.

- **Radiologist:**
  The radiologist is a specialist in medical imaging techniques (x-rays, ultrasounds, scanners and MRI). A radiologist could be called upon to help identify the diagnosis causing dwarfism, by observing the structure of the bones and skeletal irregularities. In addition, in the event of complications, it is the radiologist who can identify skeletal issues (scoliosis, kyphosis, etc.), tumors or pathology of the spinal cord. They are therefore present alongside other specialists to support little people and help them manage the health concerns associated with their condition.

- **Anesthetist:**
  Little people who are going to have an operation will need to meet with a qualified anesthesiologist. Indeed, anesthesia in people with dwarfism often requires precautions. Refer to the “anesthesia” sheet for more details on this subject.

- **Social worker:**
  Social workers offer practical support to people facing complex social realities (family problems, loss of autonomy, poverty, etc.), allowing them to find solutions to their problems. In the case of dwarfism, social workers can help little people find the resources they need.

RESOURCES:

Association québécoise des personnes de petite taille
https://www.aqppt.org/

Little People of Ontario
https://littlepeopleofontario.com/
Geneticist:
Association des médecins généticiens du Québec

Orthopedist:
Association d’orthopédie du Québec
https://www.orthoquebec.ca/la-profession/

Neurosurgeon:
Association de neurochirurgie du Québec
http://www.ancq.net/fr/la_neurochirurgie/qu_estce_que_la_neurochirurgie.asp

Physiatrist:
Institut de physiatrie du Québec

Otolaryngologist:
Association d’oto-rhino-laryngologie et de chirurgie cervicofaciale du Québec
https://orlquebec.org/

Psychologist:
Ordre des psychologues du Québec
https://www.ordrepsy.qc.ca/accueil