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Dermato-Venereological Research at Turku University

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The Department of Dermatology and Venereology at the University of Turku and Turku University Hospital was established in 1944. Over the years, research at the department has focused on proteases in cutaneous biology and diseases, allergology, fungal infections of the skin, occupational dermatoses, the proliferation of epidermal cells, and the immunosuppressive effects of sunlight. So far, 35 doctoral dissertations have been published, many in co-operation with basic science departments. At present, two research groups at the department receive support from extramural research grants: the Metalloproteinases group headed by Professor Veli-Matti Kähäri and the Cell-cell interactions and Neurofibromatosis group headed by Associate Professor Sirkku Peltonen and Professor Juha Peltonen.

Matrix metalloproteinases in the skin

The Metalloproteinases research group is located at the Department of Dermatology, Turku University Hospital and at the MediCity Research Laboratory, University of Turku. The group consists of three postdoctoral researchers, five graduate students, and two technicians.

The overall aim of the group's research is to elucidate the roles and regulation of proteinases, including matrix metalloproteinases, in the growth and progression of melanoma and non-melanoma skin cancer, and in wound repair. Our aim is to identify novel diagnostic and prognostic biomarkers for progression of cutaneous cancer. Furthermore, understanding the molecular mechanisms regulating the invasive phenotype of malignant cells is also important in the development of specific therapeutic modalities for skin cancer. The group is funded by grants from the Academy of Finland, the Sigrid Jusélius Foundation, the Finnish Cancer Research Foundation, and Turku University Hospital.

Cell-cell interactions and neurofibromatosis research

The Cell-cell interactions and Neurofibromatosis research group is located at the Department of Dermatology, Turku University Hospital and at the Institute of Biomedicine, University of Turku. The group is led by Associate Professor Sirkku Peltonen and Professor Juha Peltonen. It has two postdoctoral researchers and six PhD students, of whom four are medical doctors.

Cell-cell interactions

Cell-cell interactions are essential for the normal development, differentiation, and integrity of the epidermis. A specific type of intercellular junction, the tight junction, has recently been characterised in human epidermis. Our research group has characterized human epidermal tight junctions in normal developing and adult skin. We have also shown that various aberrations in epidermal differentiation and integrity, such as those that occur in psoriasis and acantholysis, and during wound healing, lead to abnormal expression of certain tight junction components (1–3). The regulation of tight junction components is being further elucidated at the cell signalling level.

Mutations in genes encoding desmosomal components lead to combinations of symptoms, including woolly hair, acantholysis, palmoplantar keratoderma, and cardiomyopathy. We characterised a new mutation in the desmoplakin gene in a patient with all described symptoms, and enamel abnormality as a new finding (4). The patient died prematurely as a result of cardiomyopathy (at 14 years of age).

Neurofibromatosis type 1 research

Neurofibromatosis type 1 (NF1) is a dominantly inherited neurocutaneous syndrome with an incidence of about 1:3,500. It is characterized by the appearance of neurofibromas and pigmented spots (café au lait macules) in the skin. Since NF1 is one of the most common inherited syndromes, and since the symptoms are readily visible, all dermatologists should be familiar with the diagnosis of NF1. Although the *NF1* gene was identified over 20 years ago, the function of the corresponding protein, neurofibromin, is not fully understood.

Professor Veli-Matti Kähäri, Head of the Department of Dermatology and Venereology.

Sirkku Peltonen, Associate Professor in-Dermatology.

Neurofibromin is a tumor suppressor protein which regulates the Ras pathway by accelerating the inactivation of active Ras-GTP to Ras-GDP. In neurofibromatosis, Ras signaling is overactive in various cell types, leading to aberrations in cell differentiation and proliferation.

Neurofibromatosis is being investigated at the molecular, cellular, clinical, and epidemiological levels. Our most recent findings include characterisation of neurofibroma tumour stem cells (5). We have also shown that multipotent tumour stem cells are likely to reside in hair roots in association with stem cells. The pathogenesis of NF1 was reviewed by Jouhilahti et al. (6).

During the past five years, we have carried out a large number of studies on the oral manifestations of NF1 in collaboration with researchers at the Department of Oral Diseases, Turku University Hospital (7–10). The results show that various oral symptoms, such as mucosal neurofibromas, are very common in NF1. Although they may be disturbing, patients may not mention them if not asked by their doctor. We also described a novel NF1 manifestation, periapical cement dysplasia, which was solely found in women. We further showed that at least one third of NF1 patients have speech problems (11), which may lead to more or less severe communication problems.

Osteoporosis is a common feature of NF1. We recently showed that NF1 patients have overactive osteoclasts (12).

Studies of various aspects of NF1, including bone symptoms, are ongoing. Moreover, as a new research line, we have started epidemiological studies of NF1 and cancer in Finland.

Turku University Central Hospital
White arrow indicates the position of Department of Dermato-Venereology.

Facts

Turku University was founded in 1640. It relocated to Helsinki in 1828.

The University has approximately 3,000 researchers and teachers, professors, and employees, and a total of 20,000 students. There are 7 faculties at the university. The first professor of Dermatology and Venereology at Turku University was T. E. Olin, who was appointed in 1944.

The group of researchers at the Department of Dermatology in Turku consists of:
1 Professor
2 Clinical instructors
3 Associate Professors
3 Other specialists who are active in research
7 registered PhD students

The Department produces some 10 original publications every year. During the last 5 years, 2 doctoral theses have been produced.