

## ONCHOCERCIASIS

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**Introduction.** Onchocerciasis – or “river blindness” – is a parasitic disease caused by the filarial worm *Onchocerca volvulus* transmitted by the bites of infected blackflies (*Simulium* spp.) repeatedly. These blackflies breed along flowing rivers and stream. This disease is common in Africa, and also in Latin America and Yemen. It was discovered that 17.7 million people are infected with this disease, 95% in Africa. Approximately 270,000 people are blind as a result of this disease, while others have serious visual impairment. As of 20 July 2016, transmission of the parasite of onchocerciasis is still ongoing in Brazil and Venezuela.

**Aim.** The current study focuses on the onchocerciasis infection and its origin, mode of transmission and possible cure.

**Material and methods.** Descriptive method was used based on the analysis of 13 scientific articles.

**Results and discussion.** The microfilarial parasite that causes the disease was first identified in 1874 by John O’Neill, the Irish naval surgeon. In the human body, the adult worms produce embryonic larvae (microfilariae) that migrate to the skin, eyes and other organs. When a female blackfly bites an infected person during a blood meal, it also ingests microfilariae which develop further in the blackfly and are then transmitted to the next human host during subsequent bites. Onchocerciasis is an eye and skin disease. In the early stages, the symptoms may not appear. It can take up to a year for the infections to become apparent. Symptoms are caused by the microfilariae, which move around the human body in the subcutaneous tissue and causes inflammation when they die. Infected people may show symptoms such as severe itching and various skin changes, enlarged groin, cataracts, lymph node inflammation and light sensitivity. Some infected people develop eye lesions and permanent blindness. There is no vaccine or medication to prevent infection with *O. volvulus*. The simplest and the best way to prevent being infected is personal protection measures. This includes wearing clothes that does not expose the skin and wearing insects repellent such as DEET.

**Conclusions.** WHO recommends treating onchocerciasis with ivermectin at least once yearly for between 10 to 15 years. This medication paralyzes and kills microfilariae by interfering with the nervous system and muscle function. It does not kill the adult females but stops them from releasing microfilariae.

## GENETICS AND EPIDEMIOLOGY OF THE PROTEUS SYNDROME (THE ELEPHANT MAN SYNDROME)

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**Introduction.** Proteus syndrome is a rare disorder characterized by overgrowth of various tissues of the body. Affected individuals may experience a wide variety of complications that may include progressive skeletal malformations, benign and malignant tumors, malformations of blood vessels (vascular malformations), bullous pulmonary disease, and certain skin lesions.

The **aim** of the study was to investigate the causes, population rates and detailed symptoms of the disease.

**Materials and methods.** Analysis of medical literature available.

**Results and discussion.** Proteus syndrome may affect bone and connective tissue, fatty tissues, skin, central nervous system and internal organs (viscera). The specific symptoms and severity varies greatly from case to case. In some cases, affected individuals may exhibit only a few, mild symptoms of Proteus syndrome, making diagnosis extremely difficult. Proteus syndrome is caused by a mutation in a growth regulatory gene called *AKT1* that occurs after fertilization of the embryo (somatic mutation).