

ICHTHYOSES AND ECTODERMAL DYSPLASIA

The ichthyoses are a group of inherited skin disorders that have excessive scaling as a common feature. Most of them are lifelong conditions and they vary greatly in extent and severity. Classification of the ichthyoses is based on: 1) clinical features, for example, age of onset, type and locations of scales; 2) the appearance of the skin under the microscope and 3) the inheritance pattern within the family. The biochemical and/or structural defects in the skin have been identified in a few of the disorders, and will be discovered in others as we learn more about the human genome. In virtually all forms of ichthyoses, it is the most superficial layer of the skin, the epidermis that is abnormal. This layer either proliferates more rapidly than normal, resulting in visible scales or its more superficial portion, the stratum corneum, fails to be shed normally, also resulting in visible scaling.

Ectodermal Dysplasias and ichthyoses share some features. Infants with both types of disorders can have dry, peeling skin and sparse hair at birth. The dry, scaly skin found in older children with certain types of ichthyosis also may resemble ED skin. The sweat glands are usually normally developed in ichthyosis, but may function poorly because thick scales plug the sweat pores and form a mechanical barrier to sweating. Thus, some individuals with ichthyosis may be intolerant of hot weather and, like EDs, flush and feel uncomfortable when the environmental temperatures are high. The most common type of ichthyosis, called ichthyosis vulgaris, frequently occurs in association with eczema (also known as atopic dermatitis or infantile eczema). This combination of skin disorders may mimic the findings in some EDs where the skin is dry, sensitive, and prone to developing eczema.

In the overwhelming majority of individuals with ichthyosis, the hair is normal. Occasionally, if there is severe involvement of the scalp, the hair cannot penetrate the thick scales and appears sparse and abnormal in texture. In a few of the rare ichthyosis syndromes, there are specific abnormalities of the hair shafts. These structural defects can be detected by microscopic examination of a small sample of hair. Likewise, the nails in ichthyosis are almost always normal. Rarely, they can be thickened, abnormally curved or grow excessively rapidly. Dental abnormalities are not associated with ichthyosis.

Supporting a normal lifestyle