CLASSIFICATION FOR THE ECTODERMAL DYSPLASIAS

The classification is built around proper use of medical terminology and is meant to bring some order to the ever-expanding numbers of disorders that are proposed to be included under the ED designation.

The classification is really quite simple. It starts by looking at the meaning of the two words in the ED designation: ‘ectoderm’ refers to the outer covering of the developing baby and ‘dysplasia’ refers to abnormal development. Thus, abnormal development of any single structure derived from the ectoderm (hair, nails and teeth to name a few) can be referred to as an Ectodermal Dysplasia. For instance, an inherent defect of hair can be called an Ectodermal Dysplasia (Trichodysplasia – ‘tricho’ being the prefix for hair), an inherent defect of the nails can be called an Ectodermal Dysplasia (Onychodysplasia – ‘onycho’ being the prefix for nails), and an inherent defect of the teeth can be called an Ectodermal Dysplasia (Dentodysplasia – ‘dento’ being the prefix for teeth).

This seems simple enough, but confusion enters the picture when one thinks about the historic definition of ED: a genetic disorder that has abnormalities of two or more derivatives of the ectoderm. There is no problem with the word genetic; regardless of what else one does in calling something an ectodermal dysplasia, the genetic basis is a constant. It’s the phrase two or more that’s the problem. In genetics, there is a word to indicate that two or more physical traits co-exist; it is ‘syndrome’. Thus, what people have always been calling Ectodermal Dysplasias are really syndromes that involve the ectoderm. The classification takes this into account by saying that in addition to Ectodermal Dysplasias (defects of single ectodermal structures); there are Ectodermal Dysplasia syndromes (simultaneous defects of two or more ectodermal structures).

The ED syndromes may be pure in the sense that only ectodermal structures are involved or complex when structures not derived from the ectoderm are involved. Two examples will clarify this concept. Hypohidrotic ED affects predominantly the hair, teeth and sweat glands; it is a pure ED syndrome. The EEC syndrome affects the hair, teeth, sweat glands and bone (cleft lip/palate and ectrodactyly); it is a complex ED syndrome. This is all simple enough, but what about syndromes that primarily affect non-ectodermal structures but have mild, occasional or secondary involvement of ectodermal structures? Simple, these are called related syndromes. The Trichorhinophalangeal syndrome is an example of a related syndrome; it affects the hair, but its primary features are in facial structure and bones.

Are you asking who cares? It seems like such a useless exercise, like nit-picking. The classification is important, however, for scientists, the ED Society and families. Scientists must define and separate the EDs carefully if they are going to make any headway in discovering what causes them. Without a clear classification, it’s as hard for scientists to know what is going on as it is for a child to colour a picture with a box of crayons that have melted together. The ED Society must deal with classification to know who it is serving, what their needs are and what disorders it should expend its scarce resources on. Families must deal with the classification to be able to describe to doctors exactly what disorder concerns them, to know what to expect (will the bones be all right or not?), and to communicate effectively with other families.
Even though classification is important, there is one thing that families should not forget. Like families who come to this country: we didn't all come over on the same ship, but we're all in the same boat. It's the common experiences of families that draw them together and that will lead to discoveries about causes and advances in treatment, and will ultimately lift all our collective hopes for the future.