Epidermodysplasia verruciformis type 1

GENERAL INFORMATION

Description:

Epidermodysplasia verruciformis (EV) is a rare, lifelong, autosomal recessive hereditary disorder affecting the skin.

Alternative names:

- EV1
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Classification:

- Other well-defined immunodeficiency syndromes
  - Epidermodysplasia verruciformis

Inheritance:

Autosomal recessive

OMIM:

- #226400 Epidermodysplasia verruciformis; EV
- *605828 Epidermodysplasia verruciformis gene 1; EVER1

Cross references:

Phenotype related immunodeficiencies:

- IDR factfile for Epidermodysplasia verruciformis 2

Incidence:

Incidence is not known.

CLINICAL INFORMATION

Description:

The disease is characterized by chronic infection with human papillomavirus (HPV). Widespread skin eruptions of flat-to-papillomatous, wartlike lesions and reddish brown pigmented plaques on the trunk, the hands, the upper and lower extremities, and the face are characteristic. The lesions may transform into malignant carcinomas, usually after age 30 years. Skin cancers initially appear on sun-exposed areas, such as the face and the ear lobes. Patients with EV are usually infected with multiple types of HPV. More than 30 HPV types, including types 3, 5a, 5b, 8-10, 12, 14, 15, 17, 19-21, 23-26, 37, 38, and 47, have been identified in EV tumors. EV results from an abnormal susceptibility to specific related human papillomavirus (HPV) genotypes and to the oncogenic potential of some of them, mainly HPV5. Infection with EV-associated HPV leads to the early development of disseminated flat wart-like and pityriasis versicolor-like lesions. Patients are unable to reject their lesions, and cutaneous Bowen carcinomas in situ and invasive squamous cell carcinomas develop in about half of them, mainly on sun-exposed areas. The lesions often resemble verrucae planae. The mucous membranes, hair, and nails are not affected. Malignant degeneration, usually of the superficial basal cell type, is frequent. Characteristic changes in the epidermal cells with peculiar vacuolization are observed.

Diagnosis:

Diagnostic laboratories:

Clinical:

- Lutz-Lewandowsky epidermodysplasia verruciformis, ORPHANET
- Epidermodysplasia verruciformis, eMedicine

Therapeutic options:

- Experimental therapies include intralesional administration of interferons and retinoids. These therapies have resulted in only a partial or transitory effect. In advanced HPV-related carcinomas, an experimental therapy involves treatment with a combination of 13-cis retinoic acid and interferon alpha or cholecalciferol analogues. For localized multiple malignant lesions, autotransplantation of skin from uninvolved skin has been reported with success in preventing further development of cancers. UV-B and UV-A exposure as well as x-ray irradiation should be avoided.
- Epidermodysplasia verruciformis, eMedicine

Research programs, clinical trials:

- European Initiative for Primary Immunodeficiencies 2001-2004

GENE INFORMATION

Names:

HUGO name: TMC6
Alias(es): EV1, EVIN1, LAK-4P, epidermodysplasia verruciformis 1, expressed in activated T/LAK lymphocytes, EVER1 protein (Fragment), EVER1

Localization:

Reference sequences:

DNA: BC018346 (EMBL) , **cDNA:** X58957 (EMBL) , **Protein:** Q7L2M4 (SWISSPROT)

Chromosomal Location:

17q25

Maps:

EVER1 (Map View)

Markers:

D17S939, D17S802, D17S1839, D17S1802, RH80030, D11S3114, PMC181332P1

Variations / Mutations:

- TMC6base; Mutation registry for Epidermodysplasia verruciformis (EVER1)

Other gene-based resources:

Ensembl: ENSG00000141524, GENATLAS: EVER1, GeneCard: EVER1, UniGene: 16165, Entrez Gene: 11322, euGenes: 11322, HomoloGene: 5258

PROTEIN INFORMATION

Description:

Other features:

Expression pattern for human:

Tissue	Exp. (%)	Clones
thymus	22.35	2:2217
spleen	20.07	10:12346
blood	14.00	26:46017
thymus	8.06	1:3074
lymph_node	4.79	22:113869
stomach	3.11	9:71656
bladder	3.03	2:16340
colon	3.01	10:82275
skin	2.81	17:150171
bone_marrow	2.53	3:29369

Animal models:

Mouse:

MGD: ; Tmc6, NCBI Gene: ; 217353 (76.19 % aminoacid similarity to human)

OTHER RESOURCES

Societies:

General:

- International Patient Organization for Primary Immunodeficiencies
- Immune Deficiency Foundation
- European Society for Immunodeficiencies