Epidermolysis Bullosa Heriditaria

— a brief survey and clinical data on 56 living persons with epidermolysis bullosa dystrophica dominans

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Introduction:

In a previous report the author has given a brief description of some clinical findings in two families in the Faroe Islands in which epidermolysis bullosa heriditaria (EBH) occurred and a genetical analysis was carried out[18]. Furthermore, a more precise classification was discussed. The conclusion was that the affected members in one of the families (family A) could be classified in the subgroup EBDD (Bart), while the affected members in the other (family B) were classified in the subgroup EBDD (Cockayne-Touraine).

In the first section of this paper a brief survey about EBH in general is given. Otherwise the paper primarily deals with a closer statement of the clinical findings in the affected persons in the two families.

Survey.

von Hebra[16] in 1870 described a case with familial non-scarring traumatic blistering of the skin. In 1879, *Fox*[9] directed the attention to a skin disease with traumatic blistering of the skin associated with scarring and nail dystrophies. The disease received its present common name, epidermolysis bullosa heriditaria, from *Koebner*[20] in 1886.

Classifications of cases of epidermolysis have been rather

heterogeneous, even confusing, throughout time. *Hallopeau*[15] classified the disease into two main groups: simplex and dystrophica while *Siemens*[38] classified it into a dominant (simplex) and a recessive (dystrophica) form. According to *Herlitz*[17] the dystrophic recessive form should be subdivided into two groups: dystrophic and lethalis.

On the basis of an analysis of a series comprising 1181 cases, *Touraine*[42] described three different types of EBH: two dominant types were termed epidermolysis bullosa simplex and epidermolysis bullosa hyperplastica, respectively, the third, recessive type being termed epidermolysis bullosa polydysplastica. With support in some histobiochemical studies, *Lowe*[21] classified the disease into three groups: Epidermolysis bullosa simplex, epidermolysis bullosa dystrophica, dominant form (EBDD), and epidermolysis bullosa dystrophica, recessive form (EBDR). This is in accordance with the classification of Touraine. *Love* suggested to abandon the term »lethalis« and rather apply the term EBDR, severe form, to fatal cases of epidermolysis bullosa.

Gedde-Dahl jr.[13] has recently set upp a new classification according to which three of EBDD may be differentiated, namely EBDD (Cockayne-Touraine), EBDD albopapuloidea (Pasini) and EBDD (Bart).

EBH is a rare disease. According to Scandinavian publications[4 8 12], epidermolysis bullosa is in evidence for the time being in about 100 patients in each of the countries Sweden, Norway and Denmark. Several Scandinavian families have been studied with the aim of defining the genetic characteristics involved[4 7 12 23].

A few reports on epidermolysis bullosa acquisita have appeared, but it remains open to discussion whether the cases concerned may not have been of recessive type[25 34].

Data concerning the disease in general have appeared in several publications[11 12 29 30 42]. In severe cases the disease may be responsible for contractures of fingers and hands[28 40], as well as for cicatricial lesions involving the alimentary canal[10 36],

deformities of teeth[36], severe haemorrhages from the oesophagus and rectum[36 41], hepatic and renal amyloidosis[5], and squamous-cell carcinomas has been seen after oral lesions[36]. Congenital aplasia of the feet has occasionally been encountered[3 7 24 36]. Also other congenital amomalies have been found to be more common in cases of EBH than in the normal population[11].

So far, EBH has not been found to involve chromosomal anomalies[39].

Studies of the formation of dentin and the structure of enamel indicate that EBDR may be of ectodermal origin[1 31].

It has been endeavoured in a series of biochemical and histological studies to define the aetiology and pathogenesis of the disease as well as to differentiate between the various subtypes. According to the biochemical analyses, the mechanism of blister formation, whether in cases of epidermolysis acquisita or EBDR, seems to be associated with an abnormal synthesis of collagen or a presence of abnormal collagen produced in some other way[26 27 33]. Available data indicate that distinctly different causal mechanisms operate in the different types of the disease (EBS, EBDD, and EBDR)[21 27]. By and large, there is a consensus of opinion as regards the localization of blisters in the three types of disease[21 30 35] whereas the importance of the elastic fibres for the cohesion between epidermis and dermis is widely discussed[28 30]. On certain occasions, parameters of hemostasis have been thoroughly studied in patients with epidermolysis bullosa, but conditions were found to be normal[8 14].

Several other hypotheses concerning the aetiology of the disease have been advanced. *Winer*[44] suggested that the disease might be attributable to a hereditary vascular anomaly of the skin while *Lutowiecki*[22] held a congenital defect in the hyaluronidase-hyaluronic-acid system responsible.

Reports on effects of tentative, systemic therapies are rather sporadic. Steroids, antimalarial drugs, and alpha-tocopherol have occasionally been found to be of value[2 6 32 35 43]. Local

treatment with 0.2 % solutions of fluocinolon acetonide[37] or 0.5 % solutions of silver nitrate[19] has been found to be of a certain effect in severe cases. In the management of sequelae such as contractures, plastic surgery has occasionally been applied[28 29].

Material

Generally speaking, the data have been collected personally by the author who interviewed the affected individuals in the two families during a period from June 1970 to December 1970; both families are residing in the Faroe Islands. The two genealogical tables are constructed on the basis of information thus obtained by the author from several affected as well as unaffected members of the two families. The greatest possible number of these were examined personally by the author. Occasionally it might prove impossible to have personal contacts established, if so, information was obtained via postal questionnaires. Thus, the study is primarily based on genetic characteristics and clinical symptoms.

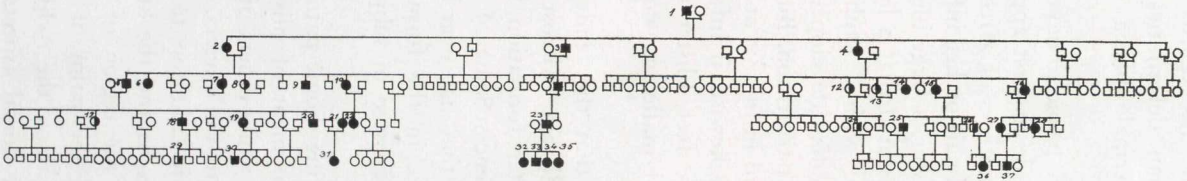
Both of the affected families originate from the largest of the Faroe Islands, Streymoy, family A coming from the northern Vestmanna district, family B from Tórshavn. A thorough study of the genealogical trees showed that the earliest ancestors in the two families were kindred (fig. 1, A and B). It will appear from the discussion why it has been preferred to discuss the two families separately.

Results:

Family A

The total number of affected individuals who are still alive is recorded in table I. It will be seen from Fig. 1 that the disease is regularly transmitted by a dominant, autosomal gene although manifestations may be of varying severity; for instance, a tendency to bullaformation was not in evidence in eight of the affected individuals. In one of the patients, a four-year-old child, the disease had not yet become manifest except

Fam. A



Fam. B

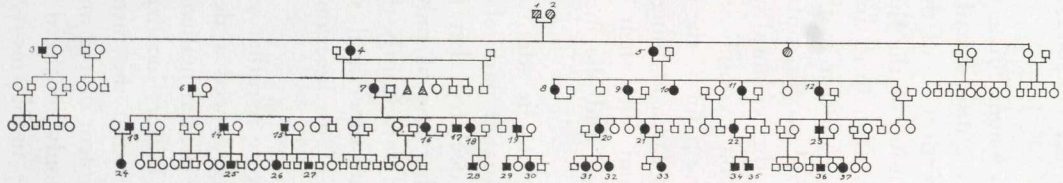


Fig 1. Occurrence of epidermolysis bullosa dystrophica dominans in the Faroe Islands.
Fam. A: Genealogical table applying to family A.
Fam. B: Genealogical table applying to family B.

□ men; ○ women; △ sex unknown; □○ healthy individuals; ■● patients in whom bullaformation occurs; ◨◑ patients in whom deformed nails is the exclusive symptom; ▨⊘ no data of morbidity.

in the form of a congenital dermal aplasia. As regards the remaining seven patients, lesions other than deformities of nails were not observed; yet congenital dermal aplasia was manifest in two.

All of the 20 subjects in whom bullae were prone to develop were of the opinion that the disease had made its first appearance at or briefly after birth. One of the mothers of affected infants declared that suffusion of the thumbnail of her infant had been observed the day after birth and later, a large blister involved the entire thumb. Another mother stated that a large fluid-containing blister, not to be confused with a kephalic haematoma, at the site of the temporal bone had developed in her infant immediately upon a complicated parturition. Bullae were seen to develop in the palm of the hand in infants at the time when they started to catch at things. Likewise, if infants were allowed to kick about with their bare feet, blister formation involving the heels and the medial malleolus would be common occurrences.

All except the small children admitted that the tendency to bulla formation had abated parallel with age. Almost all members of this family declared that blister formation had subsided completely at the time of pubescence. Some of the adult subjects remembered only faintly that they had ever had such blisters. Nothing but superficial scars, in the form of atrophic skin at the sites of predilection, persisted in subjects of middle-age.

The severity of the disease is rather varying. Some patients had only occasionally experienced a development of blisters involving the knee or the medial malleolus while others often had been suffering from blisters localized to the fingers, the elbow, the back, the shin and the instep in addition to an almost permanent involvement of the malleolus and the knee.

Bullae are most commonly localized to the region of the medial malleolus whereas involvements of the palm of the hand, the processus spinosus, the shin, sites above the Achilles tendon on the medial edge of the foot, the dorsal aspects of

toes, the theca cranii, the external meatus, and the oral cavity are less frequent.

The factors responsible for a development of blisters are in all cases mechanical traumata of different types. The most common traumata are those inflicted to ankle bones due to a jarring of the joint, and those inflicted to the knee. In infants who are kicking their naked feet, jarring of their heels will often be responsible, or they may clutch their toys with their hands; when they are lying naked on a hard bed, kicking their feet, the skin above the processus spinosus may be injured.

Usually, bullae will develop within about 10 to 20 minutes without any preceding symptoms.

In rare cases, especially in the case of infliction of parallel pressures or traumata, bullae may not develop but the epidermis will primarily, when touched, roll away like thin tissue paper.

The sizes of bullae may vary; for instance, bullae on the knee may be 10 cm in diameter, but diameters of a few mm are also seen. Most of the bullae are found to measure 1 or 2 cm across.

Eruption of bullae occurs generally within the initial 48 hours after onset. Major bullae are mainly punctured by the patients themselves because they otherwise may grow larger. They are generally seen to contain a clear, colourless fluid, but it may also be sanguinolent. The interval until the floors of bullae are healed covers mainly 10 to 14 days. Wound infection is a rare occurrence.

All patients declared that the bullae left superficial scars (Table I); the scars appeared as a thin, dry, hairless, glistening, atrophic skin in which pigmentation was absent; occasionally and depending on the length of time after appearance, their floors might be of erythematous character, reminding of scarring after mild burns.

Changes of the nails were manifest in all patients except three children of 1, 3, and 12 years, respectively. The author has not personally had occasion to examine two of these chil-

TABLE I.

Number of patients and distribution of certain characteristics.

	Family A	Family B
Total no. of patients	37	36
Total no. of living patients	28	28
Malformation of nails	25	26
Malformation of nails, exclusive symptom	7	0
Congenital dermal aplasia	6	0
Congenital dermal aplasia, exclusive symptom	1	0
Tendency to bullaformation	20	28
Scarring	20	28
Milia	12	8
Bullae involving mucous membranes	6	5
Excoration when adhesive tape is removed	3	3
Dental anomalies	1	7
Improvement during summer months	0	2
Aggravation during summer months	6	0

dren one of whom is living in Denmark, the other in Greenland. I some cases the nails were seen to become deform immediately after birth, in others the deformities might develop years later. Such deformities are of varying character, ranging from complete anychia to very slight deformities. Affected nails are often shorter, thicker, and of a more granular nature than normal ones; they are discoloured into a yellowish-brown hue. The toe nails were implied in the 25 cases of nail deformities. Affection of finger nails was less common and was seen only in 14 out of the 25 patients.

Table I illustrates the tendencies to milia, excoriation when adhesive tape was removed, congenital dermal aplasia, and bulla formation involving the mucous membranes. The congenital dermal aplasia was in all cases localized to the feet, to one or to both. Bullae of mucous membranes were encountered only in the oral cavity. The chewing of dried fish or carrots might be the releasing factor.

Dental anomalies were seen only in one patient in whom impaction of four canine teeth was observed. In five patients,

the trunk was a site of a uniform skin eruption which persisted until pubescence at which time it subsided. This eruption did not remind of bullaformation. Anomalies other than those were not encountered in any member of this family.

Six of the questioned patients declared that the tendency to bullaformation aggravated slightly during the summer months. One explanation may be that children are more lightly clad at that time of the year and consequently they are less protected, another that the plays in which children join may be of a more violent character during the summer than in the winter.

None of the patients had found the disease of invalidating nature, but all admitted that the inconveniences involved were severe. Such inconveniences were individually evaluated. Some patients might emphasize the pain and smarting sensation brought about by the denuded surfaces of wounds and, for instance, such wound surfaces on the medial malleolus might adhere to the stockings if bandages were not applied; it might therefore be painful to take off the stockings at bed time.

Mothers of affected children had to face the inconvenience of a permanent care of their wounds.

It requires unremitting attention to escape a development of blisters and deformities of nails. It is a nuisance to the children always to be aware of their disease and pay extra attention to it when they are playing and hurts are to be escaped or to be careful that nails are not caught in doors or otherwise injured. Boys may not be able to join in football on account of their tender toe nails and their sensitive malleolus and knees.

The lesions may also represent a psychic strain. One male patient, for instance, admitted that he during childhood had developed complexes because he found himself more awkward than other boys of his age in whom bullae and scars never occurred. The deformed nails may mean agonies to young girls. If they go about barefoot, people will stare at them and consequently they would for instance only reluctantly go to bathing beaches.

Bullae are mainly punctured by the patients themselves. The membrane covering the blister is rarely removed because patients have come to learn that healing will be more rapid after puncture of blisters when they remain covered by the thin film of skin and hence, they prefer this procedure to that implying removal of the skin by which the denuded wound will be more exposed to injury. Another advantage of puncture is that it arrests further growth of the blisters.

The wounds *per se* are usually treated with topical application of zink- or lapis ointments and bandages. In some cases, trypoflavin compresses may be of benefit.

Some mothers applied pads of cotton wool or foam rubber to the malleolus of their children with the aim of escaping blister formation.

In a few cases, hospitalization has been indicated and the patients concerned were exposed to various experimental therapies including application of radium plates to the nails and treatments with ypsilon-aminocaproic acid, steroid ointments, and administrations of several types of tablets of unknown content, but all measures proved futile. Some patients have found therapeutic carbon light of a certain effect.

Family B

The number of affected individuals appears from Table I. Also in this family the disease was found to be systematically transmitted by a dominant, autosomal gene (cf. Fig. 1). Bulla-formation was in evidence in all affected subjects, i. e. in contrast to findings in family A, malformation of the nails was not the exclusive symptom in any of the cases. All patients maintained that they had been suffering from the disease since infancy.

Whether or not the disease had improved parallel with age was a matter of discussion; eighteen out of the 28 patients were of the opinion that improvement had occurred while eight considered their condition unchanged. Two patients did not know. Thus, there seemed to be a faint tendency towards

improvement parallel with age. It was the general opinion that blister formation had not subsided completely after they had achieved adult age. Some of those who challenged the postulation of an improvement parallel with age admitted, however, that bulla might not occur at the same rate as before, but the reason was rather that they by now had learned to be more careful and did not join in activities implying hazards of traumata.

The medial malleolus was the site most generally exposed to bullaformation, but also the knees, fingers, and instep were commonly affected. Localizations of a more specific nature included the palm of the hand, the ulnar aspect of the wrist, sites above the acromion, the chin, the cheeks, the bridge of the nose, the shin, sites above the lowermost portion of the Achilles tendon, the lateral malleolus, the heels, the toes, and the oral mucosa.

The patients were questioned whether bullaformation might be excited by certain exogenic factors. One patient answered that bullaformation had been absent during her pregnancy, but the other women had not the same experience. Among those questioned, two had observed that bullaformation had been less frequent during the summer months, but many of the others had not been aware of such seasonal variation. Occasionally, psychic strain might be responsible for an aggravation of the condition, probably because they in such state might not be sufficiently careful to avoid traumata. Other factors such as heat, cold, humidity, alcohol, tobacco, and fever were apparently without any influence.

Infection of wound surfaces was rarely seen. In fact, many declared spontaneously that the healing capacity of their skin was extremely good.

One of the patients had noticed that bullae might be of two types, one being small contained a sanguinolent fluid and healed well spontaneously, another being larger contained a clear, serous fluid and healed best after puncture.

As regards the development of bullae, their size, aspects, and healing properties, conditions were almost the same as those observed in family A.

It was generally admitted that bullae would leave scars. In similarity with findings in family A, such scars would remind of a slightly atrophic skin.

Many patients complained that their skin in general would be of an irritating dryness.

Some of those questioned had observed that bullae to develop later than early childhood would almost always appear in scars left by those dating back to childhood.

Changes of the nails were in evidence in all of the 28 patients except two; the latter were not further examined because it had not been possible to have postal contact established except on one occasion. The toe nails were involved in all cases of deformed nails; additional involvement of the finger nails was seen in three cases. Changes of the nails are mainly due to severe traumata, for instance, if somebody happens to tread on the patient's toes,

Table I illustrates: the incidence of milia, the time of healing of the floors of bullae, excoriation when adhesive tape has to be removed, congenital dermal aplasia, and blister formation.

Dental changes were manifest in seven cases, either taking the form of discolouration and/or incorrect position of teeth; these changes were of mild degree in all cases. Anomalies other than these included: enuresis, observed in four patients; depigmented skin areas on hands and back, observed in two cases; and psoriasis, observed in one case. Besides, cleft palate was seen in one case, achylia in one, and myxoedema in one.

Most patients in family B found the disease inconveniencing. In a few of these, healing of the floors of the blisters would be associated with a certain pain and a smarting sensation round about the bullae together with itching. The most common complaint was that they always had to be careful if traumata were to be escaped. During childhood it is a severe drawback always to wear bandages or compresses. The children are

unable to join in plays with the same pleasure as other children. One of the patients had never had occasion to participate in swimming lessons because he always had open wounds.

Quite a number had felt ashamed of their disease because, on the basis of statements advanced by some doctors, they had got the impression that their disease might have arosen because one of their ancestors might have had syphilis.

Large blisters had usually been punctured by the patients themselves and the wounds treated with boiled water, boric acid solution, merbromine, zinc ointment or terramycin powder, and sterile bandages.

In a few, severe cases, treatment had included ypsilon-amino-caproid acid, vitamins A, B and C and the like, but the effects obtained were not reliably proved.

Some patients found, that carbon light had some effect.

Discussion:

As only a few of the patients had such anomalies as bullae and miliae at the time, when the author's personal examination took place, a considerable part of this investigation is built upon anamnestic information from the patients and not a personal examination. Nail deformities, scar formation, and sequelae of congenital local absence of skin could, however, be inspected in cases where the author could establish personal contact. Inspection of previously affected areas of the skin frequently failed to disclose scars. In other cases, the patients' information about scar formation could be verified, especially of the area around the medial malleolus.

It has been shown that there are 1/3 more unaffected than affected persons out of the possible in family A, even if it is clear that the disease is systematically transmitted by a dominant autosomal gene[18]. This difference could very well be accidental, but the reason could also be that the author could only establish personal contact with 50 % of the affected members of family A, and had no opportunity to examine a great number of children who were said to be non-affected in letters from their affected parents.

It applies to both families that epidermolysis bullosa makes its first appearance during infancy, but the condition may improve in adult age; either the disease subsides or the incidence of bullaformation is decreased; the tendency of bullaformation may also become less marked because the patients learn to be careful.

Inconveniences, somatic as well as psychic, caused by the disease may be considerable. It is a general trait of all patients, however, that they make the best of their condition and reconcile themselves with the fact that it is a familiar predisposition and their siblings, if any, as well as one of their parents are also affected. Only few of the patients did ever go to the doctor with the disease.

Malformation of nails represents an almost constant phenomenon in both families. In the cases without involvements of nails, the patients concerned are either infants in whom deformities later may develop or may be those not examined by the author. According to findings in the present study, malformation of nails may be the exclusive symptom of the disease in patients otherwise predisposed to a development of epidermolysis bullosa dystrophica, dominant form.

The patological pictures observed in the two families have certain features in common, for instance, the mode of development of the blisters, their content, size, localization, and healing. Milia and involvement of mucous membranes are seen in some patients in both families. It also applies to both families that the disease is systematically transmitted by a dominant, autosomal gene. Superficial scars left by bullae are seen in almost all cases. These findings suggest that both families are affected by epidermolysis bullosa heriditaria dystrophica, dominant form.

Even so, there is a certain difference between the pathological pictures observed in the two families A and B. Above all, in patients belonging to family A, bullae did never develop after the age of puberty while such formation might continue to occur in all patients belonging to family B even after they

had achieved adult age, though at a rate decreasing parallel with age. Family A comprised eight members in whom malformation of nails represented the exclusive symptom of the disease while all affected members in family B also developed bullae. It should also be mentioned that congenital, dermal aplasia was in evidence in six patients belonging to family A, whereas this symptom never occurred in members of family B.

As already mentioned, a closer placing of the affected members of the two families within the EBH-classification has been made in a preceding report.

The frequency of the disease epidermolysis bullosa dystrophica dominans in the Faroe Islands is very remarkable. We have found two dissociated families in which two types of the disease occur in 73 persons during 5 generations. If we consider the Faroe Islands, numbering a mere 40 000 inhabitants, and compare them with Europe it must surprise that this disease seems to be so rare in other countries. It is a temptation to suppose that the disease is by far more common than it has generally been thought.

SUMMARY

A brief survey of the disease epidermolysis bullosa heriditaria is given. In the Faroe Islands, epidermolysis bullosa has been found to occur in two families. Among a total of 73 affected individuals, 56 are still alive. The disease was found to be systematically transmitted by a dominant, autosomal gene. According to a review of the clinical findings in the individual cases, the disease was of dystrophic type, the symptoms including deformities of nails, scarring after bullae, miliae and, in some cases, bullae involving the mucous membranes. In one of the families, six members had congenital dermal aplasia of the feet.

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ÚRTAK

Sjúkan epidermolysis bullosa heriditaria er ein ættarbregðilig sjúka, har bløðrur á húðini, elvdar av meiðingum, eru høvuðseyðkenni sjúkunnar. Sjúkan er sjaldsom. Eftir metingum hjá øðrum finnast eini 100 fólk við sjúkuni í hvørjum av londunum Noreg, Danmark og Svøríki. Ymisk sløg finnast av hesi sjúku. Oftast verða tey býtt sundur í tríggjar aðalbólkar, nevnliga epidermolysis bullosa simplex sum arvast valdandi, epidermolysis bullosa dystrofica dominans og epidermolysis bullosa dystrofica recessiva.

Í Føroyum er sjúkan epidermolysis bullosa heriditaria funnin í tveimum ættum. 73 tilburðir vóru funnir, av hesum 28 núlivandi í hvørjari ætt. Høvundurin hevur fingið til vegar upplýsingar um tey fólk í ættunum, ið hava sjúkuna, antin — og oftast — við beinleiðis vitjan ella við brævaskifti. Báðar ættirnar eru av Streymoynni, onnur, ættin A, úr Vestmanna og hin, ættin B, úr Havn.

Á Fig. 1 sæst, at sjúkan er valdandi ættarbregðilig í báðum ættum. Í báðum ættum fingu fólk sjúkuna longu sum pinkubørn. Onnur felags eyðkenni eru, at bløðrur oftast koma á øklaknútur og knø. Bløðrur kunna eisini koma á t. d. fingrum, albogum, fótleggjum, táum og hæli. Bløðrustøddin er frá tveimum mm upp í 10 cm. Nærum øll við sjúkuni høvdu ein ella fleiri kartnegl á fótunum, summi eisini á fingrunum. Flestøll søgdu, at bløðrurnar elvdu arr.

Hóast sjúkan í ættini A líkist nógv teirri í ættini B, so er kortini munur á sjúkunum. Fyrst er at nevna, at fólk við sjúkuni í ættini A ongantíð fingu bløðrur eftir kynbúning, men sjúklingar í ættini B fingu eisini bløðrur, tá ið teir vóru vaksnir, tó í minni mun. Í ætt A vóru átta fólk, ið høvdu kartnagl sum einasta sjúkueyðkenni, men øll við sjúkuni í ættini B fingu umframt kartnagl eisini bløðrur. Harumframt vóru seks fólk í ættini A, ið vóru húðaleys á fótunum, tá ið tey vóru fødd, men hetta sjúkueyðkenni var ikki at finna hjá nøkrum í ættini B.

Niðurstøðan av hesum verður, at talan er um tvey ymisk sløg av

sjúkuni epidermolysis bullosa dystrofica dominans í Føroyum. Í aðrari ritgerð er nágreiniliga greitt frá hesum[18].

Trupulleikarnir elvdir av sjúkuni kunnu verða týðandi, bæði kropsligir og sálarligir. Kortini hava fólk við sjúkuni oftast vant seg við hana og hugsa vanliga ikki um hana. Tey vita, at sjúkan er eitt ættarbregði og at annað av foreldrunum og fleiri systkinabørn eisini hava hana. Fólk fara sjáldan til lækna við sjúkuni.

At so nógv skal vera til av sjúkuni epidermolysis bullosa heriditaria í Føroyum er ógvuliga óvanligt. Funnar eru tvær ymiskar ættir við sjúkuni. Tá ið ein hevur í huga, at í Føroyum búgva einans knappliga 40 000 fólk og vit samanbera títtleikan hjá okkum við tað, sum funnið er aðrastaðni í Europu, verður ein bilsin um, at henda sjúka skal vera so sjaldsom í øðrum londum. Ein hevur lyndi til at halda, at sjúkan aðrastaðni man vera nógv vanligari enn hildið verður.