Ege Tıp Dergisi 41 (1): 43 - 45, 2002

COEXISTENCE of POLYMYOSITIS WITH EPSDERMOLYSIS BULLOSA SIW!PLEX (Weber-Cockayne variant): CASE REPORT

POLİMİYOZİT VE EPİDERMOLİZİS BULLOZA SİMPLEX BİRLİKTELİĞİ (Veber - Cockayne varyantı): OLGU SUNUMU

Kenan AKSU[1] Gürcan KISAKOL[1] Vedat İNAL[1] Utku ATEŞ[3] Fahrettin OKSEL[1]
Gökhan KESER[1] Günseli ÖZTÜRK[4] Mehmet TÜZÜN[2] Eker DOĞANAVŞARGİL[1]

[1] Ege University Faculty of Medicine, Department of internal Medicine Division of Rheumatology, Bornova, izmir
[2] Ege University Faculty of Medicine, Department of İnternal Medicine Division of Endocrinoiogy, Bornova, izmir
[3] Ege University Faculty of Medicine, Department of Histoiogy and Embriology, Bornova, İzmir
[4] Ege University Faculty of Medicine, Department of Dermatology, Bornova, İzmir

Key Words: polymyositis, epidermolysis bullosa, Weber-Cockayne
Anahtar Sözcükler: Polimiyozit, epidermolizis bülloza, Weber-Cockayne

SUMMARY

Polymyositis, characterized with proximal muscle weakness, elevated serum muscle enzymes, myopathic findings on electromyography and muscle biopsy, is an inlammatory myopathy. Epidermolysis bullosa (EB) is a rare disease vvhich is characterized with bullae formation. We present a case of polymyositis with coexistence EB in this article.

ÖZET

Polimiyozit, proksimal kas güçsüzlüğü, kas enzimlerinde yükseklik, elektromiyografide ve kas biyopsisinde miyopatik değişiklikler ile karakterize, inflamatuvar bir miyopatidir. Epidermolizis bulloza (EB) ise, bül oluşumuyla karakterize nadir bir hastalıktır. Burada, polimiyozit ile EB'nın birlikte görüldüğü bir olgu sunulmuştur.

INTRODUCTSON

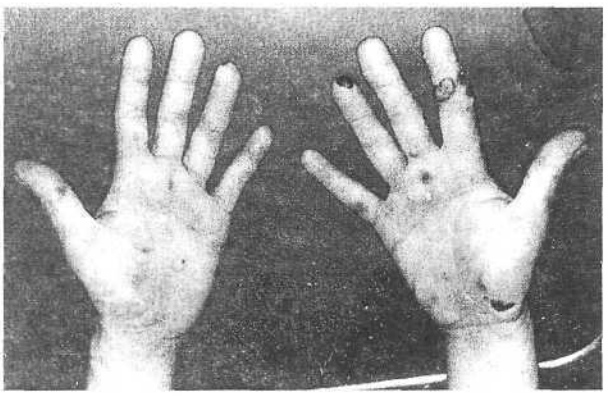
Polymyositis is an autoimmune disease primarily affecting the muscle fibers and is included within the inflammatory muscle disorders. Although muscle weakness and dysphagia are frequent symptoms reflecting the myositis, skin lesions are also observed. The most frequently reported skin lesions are pannikulitis, cutaneous mucinosis, vitiligo and multifocal lipoatrophy (1).

Epidermolysis bullosa (EB) which is characterized by formation of bullae following minör trauma of the skin has three majör subgroups: simplex, junctional and dystrophics. EB simplex which is known to be the most common subgroup has an autosomal dominant inheritance (2). Hereby we present a case of polymyositis with coexistenî EB simplex.

Yazışma adresi: Kenan Aksu, Ege Üniversitesi Tıp Fakültesi, Romatoloji Bilim Dalı, Bornova, İzmir
Makalenin geliş tarihi 20. 09. 2001; Kabul tarihi: 08. 11. 2001

CASE

A 23-year-old male with a history of bullae formation on hands and feet since childhood was referred to our clinic with the complaint of muscle weakness existing for the last 6 months. The physical examination reveaied a ioss of muscle strength by 3/5 on upper and lower proximal muscle groups. There were no accompanying Gottron's sign or heliotropic rash. Bullous lesions, prominent on the palmar sides of the hands (Figüre 1) and less on the volar side of the feet, were outstanding skin findings. The patient has two healthy siblings (one male, one femaie).



Figüre 1. Bullous lesions on the hand.

Laboratory investigations revealed ESR:32 mm/h, AST:56 U/L (5-35 U/L), ALT:79 U/L(5-40 U/L), lactate dehidrogenase (LDH) 763 U/L (230-460 U/L), creatinin phoshokinase (CPK) 2734 U/L (24-195) and aldolase 11.9 U/L (2-6 U/L). Antinuclear antibody (ANA) was positive in 1/80 titer with speckied pattern. Thyroid and surrenal functionai tests to rule out other causes of muscle weakness were within normal limits. Electromyography (EMG) revealed primary muscle involvement. Mononuclear celi infiltration, degeneration of muscle fibers and increase in connective tissue within endomysium and perimysiurn were the findings of the muscle biopsy performed afterwards. The patient was diagnosed as polymyositis with proximal muscie weakness, increase in muscle enzymes, and typical electromyographic and histopathologic findings. Radiologic imaging methods, such as ultrasonography and computerized tomography were performed in order to exclude possible coexisting malignancy.

The diagnosis of EB was confirmed by the skin biopsy, showing the separation of basal celi layer from basal membrane. Transmission electron microscopy was performed to discriminate the EB subgroups. Electron microscopic findings of the bullous skin specimen included cytolysis within basal ceils, disorganization of tonofilaments, degeneration of basal lamina and hemidesmosomes. EB simplex (Weber-Cockayne variant) was the final diagnosis confirmed with these further assays.

Treatment with oral methylprednisolone 1 mg/kg/day was started besides alendronate 10 mg/day, calcium 1.5 g/day and calcitriol 0.25 |iq/day because of the decreased bone density (lomber T score:-3.37, Z score >3.08, femur T score:-2.36, Z score:-1.91) observed by dual bone dansitometric study. The patient's osieoporosis was evaluated as occurred secondary to polymyositis.

A striking decrease of muscle enyzme levels (CPK:183 U/L, LDH:337 U/L, AST: 17 U/L) and increase in muscle strength (up to 4/5) were observed after the first two weeks of the treatment. After the normalization of the serum muscle enzymes, steroid dose was tapered gradually and oral methotrexate (MTX) 15 mg/week, was added to the treatment, with ongoing favorable response.

The patient has been in remission for two years with the current treatment of MTX 15 mg/week, methyl prednisolon 8 mg/day, anti-osteoporotic treatment and muscle strength exercises. There hasn't been any change with the above defined treatment in the skin lesions.

DISCUSSION

Inflammatory myopathies are characterized with proximal muscle weakness, elevated serum muscle enzymes, myopathic findings on EMG, and inflammatory infiltration and necrosis of the skeletal muscle on muscle biopsy. The most common inflammatory myopathies are polymyositis and dermatomyositis.

EB simplex is a rare disease with an incidence of $28.6/10^6$ (2). The clinical subgroup of EB simplex is also known as Weber-Cockayne variant. This subgroup is characterized with painful bullae formation on hands, following minör trauma or heat, starting from childhood. A minority of patients experience bullae formation on other sides of the body as well. However, oral mucosa is usually spared. There hasn't been any satisfactory treatment for EB simplex. Local treatment agents are suggested in order to prevent infections.

EB simplex was also reported to accompany muscular dystrophy (3). These patients usually suffer extensive bullous lesions, dystrophy of nails and respiratory problems. Our patient had bullous lesions restricted to hands and feet, and he had no problems with nails or respiratory system. Mononuclear celi infiltration observed on muscle biopsy, ANA positivity and good response to immunosuppressive treatment were evidences favoring inflammatory etiology of myositis in our patient.

İn literatüre, there are reported connective tissue diseases like systemic lupus erythematosus and scleroderma with epidermolysis bullosa (4,5). But, to our knowledge there is no patient reported with polymyositis accompanied by EB. This patient is the first reported case of polymyositis accompanied with EB simplex.

REFERENCES

1. Medsger TA, Oddis CV. Inflammatory muscle disease, clinical features. Klippel JH, Dieppe PA, eds. Rheumatology : 2nd ed, vol 2, Mosby, London, 1998;13:1-14.
2. Wojnarowska F, Eady RAJ, Burge SM. Bullous eruptions, congenital and inherited disorders. Champion RH, Burton JL, Burns DA, Breathnach SM, eds. Textbook of Dermatology. 6th ed. Vol 3, 1998;1817-45.
3. Shimizu H, Takizawa Y, Pulkkinen L, Murata S, et al. Epidermolysis bullosa simplex associated with muscular dystrophy: Phenotype-genotype correlations and review of the literatüre. J Am Acad Dermatol 1999;41:950-6.
4. Mc Henry PM, Dagg JH, Tidman MJ, Lever RS. Epidermolysis bullosa acquisita occuring in association with systemic lupus erythematosus. Clin Exp Dermatol, 1993; 18:378-80.
5. Ishikawa O, Warita S, Ohnishi K, Miyachi Y. A scleroderma-like variant of recessive dystrophic epidermolysis bullosa ? Br J Dermatol 1993; 129:602-5.