OBJECTIVES: To highlight the overlap between the clinical and serological features of adult Refsum disease with those of a connective tissue disease, in order to achieve the correct diagnosis and make appropriate therapeutic decisions.

Methods: Clinical and laboratory data were evaluated for 2 patients with adult Refsum disease. A medical history revealed that both reported progressive nyctalopia, anosmia, ageusia, peripheral vision loss. In addition to these clinical features one of them reported muscle weakness, weight loss and fatigue, displaying peripheral oedema, heliotrope rash, periungual erythema. Her muscular enzymes were elevated (creatine kinase was 3 times elevated and lactate dehydrogenase was 9 times above normal range), along with CRP (CRP = 2.4mg/dl). Due to clinical features, increased serum enzymes and a recently diagnosed myocarditis, a high suspicion of dermatomyositis was raised, but was disregarded after extensive investigations (immunology tests, muscular biopsy). The patient had, in addition, sicca features (dry mouth, eyes, skin) with a Schirmer test of 4mm and an elevated CRP (3.23mg/dl). She was referred to the rheumatology department with a high suspicion of Sjogren’s syndrome. Immunology and minor salivary gland biopsy were negative. Interestingly, physical examination showed bilateral brachymetacarpia and brachymetatarsia in both patients, suggesting pseudopseudoarthrodysplasia.

Results: Both patients were females in their 50s with undiagnosed adult Refsum disease. A medical history revealed that both reported progressive nyctalopia, anosmia, ageusia, peripheral vision loss. In addition to these clinical features one of them reported muscle weakness, weight loss and fatigue, displaying peripheral oedema, heliotrope rash, periungual erythema. Her muscular enzymes were elevated (creatine kinase was 3 times elevated and lactate dehydrogenase was 9 times above normal range), along with CRP (CRP = 2.4mg/dl). Due to clinical features, increased serum enzymes and a recently diagnosed myocarditis, a high suspicion of dermatomyositis was raised, but was disregarded after extensive investigations (immunology tests, muscular biopsy). The patient had, in addition, sicca features (dry mouth, eyes, skin) with a Schirmer test of 4mm and an elevated CRP (3.23mg/dl). She was referred to the rheumatology department with a high suspicion of Sjogren’s syndrome. Immunology and minor salivary gland biopsy were negative. Interestingly, physical examination showed bilateral brachymetacarpia and brachymetatarsia in both patients, suggesting pseudopseudoarthrodysplasia but, after investigations, the diagnosis was excluded. Both patients underwent ophthalmological examination to further investigate progressive nyctalopia and peripheral vision loss. Specific changes for pigmented retinopathy were described, which, together with the above neurological changes and skeletal deformities, were highly suggestive of adult Refsum disease (2). Plasma phytanic acid was 41 and 42 times within normal range (3).

Conclusion: High levels of phytic acid have triggered a series of uncommon symptoms and serological changes resembling a connective tissue disease, making the diagnosis of adult Refsum disease a very tangled, costly and lengthy process. Although this disease affects many body systems, a case of myositis or sicca syndrome hasn’t yet been acknowledged in relation to this condition.

REFERENCES

Disclosure of Interests: None declared