Abstract

Cone disorders (CDs) are a group of inherited diseases of the cone, or cone and rod photoreceptors,or retinal pigment epithelium,that are associated with various forms of stationary or progressive visual impairment.
They include achromatopsia (ACHM), cone dystrophy (COD),cone-rod dystrophy (CRD), color vision impairment, Stargardt disease (STGD) and other maculopathies.Non-syndromic inherited CDs are pathogenetically associated with forty-two genes.
Cone disorders present with a variety of clinical findings, including reduced visual acuity, photophobia and abnormal color vision.
The main diagnostic tools that can be used, except for the usual tests of visual acuity, colour vision and visual field testing are full-field electroretinogam(ffERG), fundus and near-infrared autofluorescence, optical coherence tomography(OCT) and spectral-domain optical coherence tomography(SD-OCT).
As far as cone disorders' treatment is concerned, gene therapy,molecular diagnosis and cell replacement therapies are useful tools,whereas there isn't currently any specific therapeutic plan preventing the evolution of cone and conerod dystrophies.The visual prognosis is generally poor, even though some treatment choices have been referred, includingred filters and intravitreal injection of autologous bone marrow-derived mononuclear cells.
Key words: cone disorders, genetic mutations, syndromic cone disorders, photophobia, scotoma, electroretinogram, spectral-domain optical coherence tomography, gene therapy, molecular diagnostics.