

Table title : Main clinical signs at admission of patients diagnosed with ROSAH syndrome

Ophthalmological signs						Systemic signs				Biological results					
Patient Gender age	Family history Papillary edema Uveitis	BCVA RE LE	Fundus	Posterior pole Auto Fluorescence	Macular OCT	Visual field	Recurrent fever	Spleno megaly	Severe EBV infection history	Anhidrosis	Blood count	Blood smear	Anti nuclear antibodies	Others auto anti bodies	Treatments Clinical response
Family 1 Proband M 11 yo	present	20/200 20/80	Extensive papillary edema	Hyperauto Fluorescent ring surrounding a large hypoauto fluorescent macular area	Macular edema changing towards atrophy	Coeco central scotoma	Present	present	present	present	Low platelet count 100000/mm3	poikilocytosis hyper segmented polynuclear cells	Present 1/2560 DFS +70 aspect	negative	Prednisone Colchicine Azathioprine GC intravitrealinj No clinical effects
Family 1 Proband Brother M 6 yo	present	20/20 20/20	Extensive papillary edema	Hyperauto Fluorescent ring surrounding a large hypoauto fluorescent macular area	Macular edema changing towards atrophy	Enlarged Mariotte spot changing towards coeco central scotoma	present	present	absent	present	Low platelet count 100000/mm3	NA ²	Present 1/160	negative	Prednisone Colchicine Mycophenolate mofetil Adalimumab :allergic reaction : stop Anakinra worsened clinical conditions Infliximab: +azathioprine+prednisone 10 : partial response on arthralgia No ophthalmological response
Family 1 Proband Father M 50 yo	present	1/200 1/100	Extensive papillary edema	large hypoauto fluorescent macular area	Macular edema changing towards atrophy	NA	present	present	ND ³	present	platelets 100.000-150.000/mm3 Leukopenia	anisocytosis anisochromia, poikilocytosis hyper segmented polynuclear cells	ND	negative	Prednisone for 16 years No clinical systemic benefit Worsening clinical signs
Family 2 Proband M 14 yo	present	20/20 1/200	Extensive papillary edema	Hyperauto Fluorescent ring surrounding papillar edema	Macular edema in LE	Enlarged Mariotte spot in RE NA in LE	present	present	absent	present	Low count in normal range	poikilocytosis	1/160	negative	Prednisone Anakinra for 5 months Adalimumab and MTX for 32 months No clinical and ophthalmological benefit Only prednisone 40mg/d showed improvedretinal vascular leakage
Family 2 Proband Father M 43 yo	present	20/20 20/20	Extensive papillary edema	Hyperauto Fluorescent ring surrounding papillar edema	normal	Enlarged Mariotte spot	present	Present splenectomy	present	present	normal	Howell-Jolly corpus	negative	negative	Anakinra efficient on arthralgia Canakinumab inefficient on arthralgia No ophthalmological change