Case Study 1

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AVISE™ CTD
Powered by CB-CAPs Technology

A 39 year-old Caucasian female patient first saw Dr. Goldman in November 2013 with the chief complaint of Rheumatoid Arthritis and multiple painful nodules. Based on chart notes, the patient began noting symptoms including joint pain, stiffness, deformities, muscle weakness and finger color change in cold weather back in the summer of 2004. She has a history of positive ANA, Rheumatoid Factor, CCP and Raynaud's in her teens and was diagnosed with undifferentiated connective tissue disease. Her family history consists of hypertension, high cholesterol, arthritis, cancer and osteoporosis. The patient had been on a variety of medications, including methotrexate, sulfasalazine, leflunomide (Arava), infliximab (Remicade), certolizumab (Cimzia), and hydroxychloroquine (Plaquenil). She did not respond to sulfasalazine, leflunomide and infliximab and was intolerant to some of the DMARDs. Dr. Goldman put the patient on tocilizumab (Actemra) SQ during this time, but it didn't resolve all of the symptoms, per the patient.

Over the course of 8 months and 2 follow up visits, the patient remained symptomatic with complaints of swollen hands, ankles and other joints, problems with thinking and increasing nodule pain. As a result, **Dr. Goldman ordered Avise CTD and Avise SLE Prognostic to confirm that the patient truly had overlap disease, identify which diseases were involved and determine if there could be a risk for organ involvement.**

The results of the Avise CTD showed positive markers of ANA by ELISA and IIF with a speckled pattern, anti-U1RNP, anti-RNP70, RF IgM, anti-CCP, anti-MCV and anti-thyroid peroxidase. The Avise SLE Prognostic markers were negative and the Cell-Bound Complement Activation Products markers – EC4D and BC4D – while negative were borderline, which could indicate developing SLE.

POSITIVE MARKERS	RESULTS	REFERENCE RANGE
ANA by IIF	1:640 (Speckled)	Negative (<1:80); Positive (≥1:80)
ANA by ELISA	107 Units	<20 (Negative); 20-59 (Positive); ≥60 (Strong Positive)
Anti-U1RNP	108 U/mL	<5 (Negative); 5-10 (Equivocal); >10 (Positive)
Anti-RNP70	102 U/mL	<7 (Negative); 7-10 (Equivocal); >10 (Positive)
Rheumatoid Factor IgM	91 U/mL	<3.5 (Negative); 3.5-5 (Equivocal); >5 (Positive)
Anti-CCP	170 U/mL	<7 (Negative); 7-10 (Equivocal); >10 (Positive)
Anti-MCV	138 U/mL	<20 (Negative); ≥20-70 (Positive); >70 (Strong Positive)
Anti-Thyroid Peroxidase	535 IU/mL	<60 (Negative); 60-100 (Equivocal); >100 (Positive)

The Avise results, along with the clinical assessment, provided Dr. Goldman with the information necessary to make a confident diagnosis of MCTD consisting of Rheumatoid Arthritis, potential SLE/ Lupus overlap, and Hashimoto's Thyroiditis. He was also able to rule-out internal organ involvement. Her disease activity measures have remained high: Rapid 3 = 9.3, DAS28CRP = 4.19, DAS28 ERS = 5.35, CDAI = 39 and SDAI = 39.45. Her VECTRA DA is high at 51. Based on this information, Dr. Goldman made adjustments to her current medications – Prednisone 1 mg QD, Folic Acid 1 mg QD, methotrexate 20 mg QW, probiotic QD, daily vitamin QD and Actemra SQ 162 mg/0.9 ml Q2W and requested a follow up in 3 months to evaluate her status.

> "I wanted to be certain that this patient had MCTD and identify which diseases were involved so I ordered Avise CTD. This test is unique in that it provides 2 RNP ENAs in one single test – I can't get that anywhere else!"