Fake AD Patient 7 – Young Woman with Down’s Syndrome

<Practice>

 <practiceName>UNMC Physicians<practiceName>

 <practiceAddress>

 <streetAddress>3604 Summit Plaza Dr.</streetAddress>

 <postalCode>68805</postalCode>

 <locality>Bellevue</locality>

 <region>Nebraska</region>

 <country>US</country>

 <timeZone>-6GMT</timeZone>

 <phoneNumber>14025952275</phoneNumber>

 </practiceAddress>

 <primaryCarePhysician>

 <fullName>Michael Matthews</fullName>

 <givenName>Michael</givenName>

 <familyName>Matthews</familyName>

 <license>111117</license>

 (DEA Number AJ 3274658)

 <gender>male</gender>

 </primaryCarePhysician>

</Practice>

<Demographics>

 <dateOfBirth>1978-01-31</dateOfBirth>

 <dateOfDeath>XXXX-XX-XX</dateOfDeath>

 <gender>Female</gender>

 <ethnicity>Caucasian</ethnicity>

 <language>EN</language>

 <maritalStatus>Single</maritalStatus>

 <employmentStatus>Not in work force</employmentStatus>

 <employmentIndustry> </employmentIndustry>

 <occupation> </occupation>

 <religion>Christian</religion>

 <income>5,000</income>

 <highestEducation></highestEducation>

 <organDonor>false</organDonor>

</Demographics>

<Contact>

 <name>

 <fullName>Julianne Sarah Christopherson</fullName>

 <givenName>Julianne</givenName>

 <familyName>Christopherson</familyName>

 </name>

 <email type="personal"></email>

 <email type="work"></email>

 <address type="home">

 <streetAddress>1207 E. Haven Street</streetAddress>

 <postalCode>68805</postalCode>

 <locality>Bellevue</locality>

 <region>Nebraska</region>

 <country>US</country>

 <timeZone>-6GMT</timeZone>

 </address>

 <location type="home">

 <latitude>41N</latitude>

 <longitude>95W</longitude>

 </location>

 <phoneNumber type="home">4028314040</phoneNumber>

 <phoneNumber type="work"></phoneNumber>

 <instantMessengerName protocol="aim"></instantMessengerName>

</Contact>

 <prescriptions></prescriptions>

 <conditions>

 <condition>

 <diagnosedWith>Down Syndrome</diagnosedWith>

 <dateDiagnosed>1978-01-31</dateDiagnosed>

 <diagnosedBy>Michael Matthews</diagnosedBy>

 </condition>

 <condition>

 <diagnosedWith>Alzheimer’s Disease</diagnosedWith>

 <dateDiagnosed>2009-02-01</dateDiagnosed>

 <diagnosedBy>Michael Matthews</diagnosedBy>

 </condition>

 </conditions>

 <procedures>

 <procedure>

 <procedureName>tonsilectomy</procedureName>

 <procedureDate>1986-05-22</procedureDate>

 <referredBy>Michael Matthews</referredBy>

 <referredInstitution>Bellevue Medical Center</referredInstitution>

 </procedure>

 </procedures>

 <allergies>

 <drugs>none<drugs/>

 <food>peanut butter</food>

 <environment>dust</environment>

 </allergies>

 <immunizations>

 <measles>

 <date>1978</date>

 <method> intramuscular</method>

 <compound>single</compound>

 <stage>1</stage>

 <administeredBy>Michael Matthews</administeredBy>

 </measles>

 <mumps>

 <date>1978</date>

 <method>intramuscular</method>

 <compound>single</compound>

 <stage>1</stage>

 <administeredBy>Michael Matthews</administeredBy>

 </mumps>

 <rubella>

 <date>1978</date>

 <method>intramuscular</method>

 <compound>single</compound>

 <stage>1</stage>

 <administeredBy>Michael Matthews</administeredBy>

 </rubella>

 <tetanus>

 <date>2008</date>

 <method>intramuscular</method>

 <compound>single</compound>

 <stage>1</stage>

 <administeredBy>Michael Matthews</administeredBy>

 </tetanus>

 </immunization>

<lifestyle>

 <alcoholConsumption>

 <frequency>0</frequency>

 <units>week</units>

 <quantity>0</quantity>

 <date></date>

 </alcoholConsumption>

 <exercise>

 <frequency>3</frequency>

 <units>week</units>

 <averageDuration>20</averageDuration>

 <date>2009-0201</date>

 </exercise>

 <smoking>

 <frequency>0</frequency>

 <units>week</units>

 <quantity>0</quantity>

 <history>never</history>

 <date>2009-02-01</date>

 </smoking>

 <diet>

 <fruitVegetablesRating>5</fruitVegetablesRating>

 <wholeGrainsRating>3</wholeGrainsRating>

 <dairyRating>2</dairyRating>

 <proteinRating>3</proteinRating>

 <fatsRating>4</fatsRating>

 <junkFoodRating>2</junkFoodRating>

 <date>2009-02-01</date>

 </diet>

 </lifestyle>

 <familyHistory>

 <subject\_id=”1”>

 <relation>maternalGrandmother</relation>

 <fname>Betty</fname>

 <sex>Female</sex>

 <alive>no</alive>

 <ageAtDeath>88</ageAtDeath>

 <cancer>no</cancer>

 <causeOfDeath>Old Age</causeOfDeath>

 </subject\_id>

 <subject\_id=”2”>

 <relation>maternalGrandfather</relation>

 <fname>Bob</fname>

 <sex>Male</sex>

 <alive>no</alive>

 <ageAtDeath>82</ageAtDeath>

 <cancer>no</cancer>

 <causeOfDeath>Old Age</causeOfDeath>

 </subject\_id>

 <subject\_id=”3”>

 <relation>paternalGrandmother</relation>

 <fname>Sally</fname>

 <sex>Female</sex>

 <alive>yes</alive>

 <ageAtDeath>67</ageAtDeath>

 <cancer>no</cancer>

 <causeOfDeath>Myocardial Infarction</causeOfDeath>

 </subject\_id>

 <subject\_id=”4”>

 <relation>paternalGrandfather</relation>

 <fname>Sam</fname>

 <sex>Male</sex>

 <alive>yes</alive>

 <ageAtDeath>55</ageAtDeath>

 <cancer>no</cancer>

 <causeOfDeath>Colorectal Cancer</causeOfDeath>

 </subject\_id>

 <subject\_id=”5”>

 <relation>mother</relation>

 <fname>Shirley</fname>

 <sex>Female</sex>

 <alive>yes</alive>

 <cancer>no</cancer>

 <age>73</age>

 </subject\_id>

 <subject\_id=”6”>

 <relation>father</relation>

 <fname>Stanley</fname>

 <sex>Male</sex>

 <alive>no</alive>

 <ageAtDeath>55</ageAtDeath>

 <causeOfDeath>lung cancer</causeOfDeath>

 </subject\_id>

 <subject\_id=”7”>

 <relation>sister</relation>

 <fname>Elizabeth</fname>

 <sex>Female</sex>

 <alive>yes</alive>

 <cancer>no</cancer>

 </subject\_id>

 </subject\_id>

 <subject\_id=”8”>

 <relation>sister</relation>

 <fname>MarySue</fname>

 <sex>Female</sex>

 <alive>yes</alive>

 <cancer>no</cancer>

 </subject\_id>

</familyHistory>

<Encounter>

 <dateOfEncounter>2008-11-15</dateOfEncounter>

 <physician>Michael Matthews</physician>

 <medicalBaseline>true</medicalBaseline>

 <medicalTest>Annual Medical</medicalTest>

 <reportedSymptoms>true</reportedSymptoms>

 <workingDiagnosis>true</workingDiagnosis>

 <referral>false<referral/>

 <reportedSymtoms>none</reportedSymptoms>

 <physicalObervations>

 <bodyFat%>

 <%bodyFat units=”percent”>24</%bodyFat>

 <weight units=”pounds”>130</weight>

 <height units=”inches”>60</height>

 <BMI units=”kg/m^2”>25</BMI>

 </bodyFat%>

 <waistToHips/>

 <waist units=”inches”>33</waist>

 <hips units=”inches”>42</hips>

 <waistToHip>0.8</waistToHips>

 </waistToHips/>

 <bloodPressure>

 <systolic units=”mmHg”>125</systolic>

 <diastolic units=”mmHg”>78</diastolic>

 </bloodPressure>

 <restingHeartRate>

 <restingHeartRate units=”BPM”>80</restingHeartRate>

 </restingHeartRate>

 <stamina>

 <finalHeartRate units=”BPM”>135</finalHeartRate>

 <predictedVO2Max units=”ml/kg/min”>49.2</predictedVO2Max>

 </stamina>

 <lungFunction>

 <PEFR units=”L/min”>560</PEFR>

 </lungFunction>

 </physicalObservations>

 <test = “Laboratory”>

 <CPT>85025</CPT>

 <testName>Blood count; complete (CBC), automated (Hgb, Hct, RBC, WBC and platelet count) and automated differential WBC count</testName>

 <referredName>Baljit Gupta</referredName>

 <referredClinic>Eastside Health Laboratories</referredClinic>

 <referredBy>Michael Matthews</referredBy>

 <CompleteBloodCount CPT=”85025”>

 <testCode = “HGB”>

 <CPT>85018</CPT>

 <LOINC></LOINC>

 <description>Hemogloblin</description>

 <value>13.3</value>

 <units>g/dL</units>

 </testCode>

 <testCode = “RBC”>

 <CPT>85041</CPT>

 <LOINC></LOINC>

 <description>Red Blood Cell Count</description>

 <value>5</value>

 <units>mil/mcl</units>

 </testCode>

 <testCode = “MCV”>

 <CPT></CPT>

 <LOINC></LOINC>

 <description>Mean Cell Volume</description>

 <value>92</value>

 <units>fl</units>

 </testCode>

 <testCode = “MCH”>

 <CPT></CPT>

 <LOINC></LOINC>

 <description>Mean Corpuscular Hemoglobin</description>

 <value>30</value>

 <units>pg/cell</units>

 </testCode>

 <testCode = “ART”>

 <CPT>85045</CPT>

 <LOINC></LOINC>

 <description>Reticulocyte Count</description>

 <value>1</value>

 <units>as a percent of red cells</units>

 </testCode>

 <testCode = “ESR”>

 <CPT>85652</CPT>

 <LOINC></LOINC>

 <description>Erythrocyte Sed Rate</description>

 <value>20</value>

 <units></units>

 </testCode>

 <testCode = “WBC”>

 <CPT>85048</CPT>

 <LOINC></LOINC>

 <description>White Blood Cell Count</description>

 <value>12</value>

 <units>K/mcL</units>

 </testCode>

 <testCode = “ANEUT”>

 <CPT></CPT>

 <LOINC></LOINC>

 <description>Absolute Neutrophil Count</description>

 <value>8</value>

 <units>K/mcL</units>

 </testCode>

 <testCode = “ALYMS”>

 <CPT></CPT>

 <LOINC></LOINC>

 <description>Absolute Lymphocyte Count</description>

 <value>3</value>

 <units>K/mcL</units>

 </testCode>

 <testCode = “AMON”>

 <CPT></CPT>

 <LOINC></LOINC>

 <description>Absolute Monocyte Count</description>

 <value>0.7</value>

 <units>K/mcL</units>

 </testCode>

 <testCode = “AEOS”>

 <CPT></CPT>

 <LOINC></LOINC>

 <description>Absolute Eosinophil Count</description>

 <value>0.2</value>

 <units>K/mcL</units>

 </testCode>

 <testCode = “ABOS”>

 <CPT></CPT>

 <LOINC></LOINC>

 <description>Absolute Basophil Count</description>

 <value>0.1</value>

 <units>K/mcL</units>

 </testCode>

 </CompleteBloodCount>

 </test>

 <test = “Laboratory”>

 <CPT>80061</CPT>

 <testName>Lipid panel</testName>

 <referredName>Baljit Gupta</referredName>

 <referredClinic>Eastside Health Laboratories</referredClinic>

 <referredBy>Michael Matthews</referredBy>

 <LipidPanel CPT=”80061”>

 <testCode = “LDL”>

 <CPT>83721</CPT>

 <LOINC></<LOINC>

 <description> Low-density lipoprotein</description>

 <value>4</value>

 <units>mmol/L</units>

 </testCode>

 <testCode = “TRIG”>

 <CPT>84478</CPT>

 <LOINC></LOINC>

 <description> Triglycerides</description>

 <value>2.6</value>

 <units>mmol/L</units>

 </testCode>

 <testCode = “CHO”>

 <CPT>82465</CPT>

 <LOINC></LOINC>

 <description> Triglycerides</description>

 <value>5</value>

 <units>mmol/L</units>

 </testCode>

 <testCode = “HDL”>

 <CPT>83718</CPT>

 <LOINC></LOINC>

 <description> Triglycerides</description>

 <value>1</value>

 <units>mmol/L</units>

 </testCode>

 </LipidPanel>

 </test>

 <test = “Laboratory”>

 <CPT>80076</CPT>

 <testName>Hepatic function panel</testName>

 <referredName>Baljit Gupta</referredName>

 <referredClinic>Eastside Health Laboratories</referredClinic>

 <referredBy>Michael Matthews</referredBy>

 <HepaticFunctionPanel CPT=”80076”>

 <testCode = “TP”>

 <CPT>84155</CPT>

 <LOINC></LOINC>

 <description>Serum Total Protein</description>

 <value>62</value>

 <units>g/L</units>

 </testCode>

 <testCode = “AST”>

 <CPT>84450</CPT>

 <LOINC></LOINC>

 <description>Serum glutamic-oxaloacetic transaminase</description>

 <value>88</value>

 <units>IU/l</units>

 </testCode>

 <testCode = “CK”>

 <CPT>82550</CPT>

 <LOINC></LOINC>

 <description>Creatinine kinase</description>

 <value>66</value>

 <units>U/L</units>

 </testCode>

 <testCode = “LDH”>

 <CPT>83615</CPT>

 <LOINC></LOINC>

 <description>Lactate dehydrogenase</description>

 <value>222</value>

 <units>U/L</units>

 </testCode>

 <testCode = “AMY”>

 <CPT>82150</CPT>

 <LOINC></LOINC>

 <description>Amylase</description>

 <value>144</value>

 <units>U/L</units>

 </testCode>

 <testCode = “TBIL”>

 <CPT>82247</CPT>

 <LOINC></LOINC>

 <description>Serum Bilirubin, Total</description>

 <value>20</value>

 <units>umol/L</units>

 </testCode>

 <testCode = “DBL”>

 <CPT>82248</CPT>

 <LOINC></LOINC>

 <description>Bilirubin, Direct</description>

 <value>17</value>

 <units>umol/L</units>

 </testCode>

 <testCode = “ALT”>

 <CPT>84460</CPT>

 <LOINC></LOINC>

 <description>Alanine transaminase</description>

 <value>22</value>

 <units>U/L</units>

 </testCode>

 <testCode = “ALP”>

 <CPT>84075</CPT>

 <LOINC></LOINC>

 <description>Serum Alkaline Phosphatase</description>

 <value>122</value>

 <units>IU/l</units>

 </testCode>

 <testCode = “GGT”>

 <CPT>82977</CPT>

 <LOINC></LOINC>

 <description>Serum Gamma Glutamyl Transferase</description>

 <value>242</value>

 <units>IU/l</units>

 </testCode>

 <testCode = “CRP”>

 <CPT>86140</CPT>

 <LOINC></LOINC>

 <description>Serum C-Reactive Protein</description>

 <value>66</value>

 <units>mg/L</units>

 </testCode>

 </HepaticFunctionPanel>

 </test>

 <test = “Laboratory”>

 <CPT>80069</CPT>

 <testName>Renal function panel</testName>

 <referredName>Baljit Gupta</referredName>

 <referredClinic>Eastside Health Laboratories</referredClinic>

 <referredBy>Michael Matthews</referredBy>

 <RenalFunctionPanel CPT=”80069”>

 <testCode = “NA”>

 <CPT>84295</CPT>

 <LOINC></LOINC>

 <description>Serum Sodium</description>

 <value>133</value>

 <units>mmol/l</units>

 </testCode>

 <testCode = “K”>

 <CPT>84132</CPT>

 <LOINC></LOINC>

 <description>Serum Potassium</description>

 <value>4</value>

 <units>mmol/l</units>

 </testCode>

 <testCode = “CREA”>

 <CPT>82565</CPT>

 <LOINC></LOINC>

 <description>Serum Creatinine</description>

 <value>111</value>

 <units>umol/L</units>

 </testCode>

 <testCode = “ALB”>

 <CPT>82040</CPT>

 <LOINC></LOINC>

 <description>Serum Albumin</description>

 <value>39</value>

 <units>g/L</units>

 </testCode>

 <testCode = “GLU”>

 <CPT>82945</CPT>

 <LOINC></LOINC>

 <description>Glucose Urine Test</description>

 <value>7</value>

 <units>mmol/L</units>

 </testCode>

 <testCode = “CA”>

 <CPT>82310</CPT>

 <LOINC></LOINC>

 <description>Serum Calcium</description>

 <value>2.23</value>

 <units>mmol/L</units>

 </testCode>

 <testCode = “CAI”>

 <CPT>82330</CPT>

 <LOINC></LOINC>

 <description>Ionized Calcium, Serum/description>

 <value>1.01</value>

 <units>mmol/L</units>

 </testCode>

 <testCode = “BUN”>

 <CPT>84520</CPT>

 <LOINC></LOINC>

 <description>Blood Urea Nitrogen</description>

 <value>5</value>

 <units>umol/L</units>

 </testCode>

 </RenalFunctionPanel>

 </test>

 <test = “Laboratory”>

 <CPT>84443</CPT>

 <testName>Thyroid stimulating hormone (TSH)</testName>

 <referredName>Baljit Gupta</referredName>

 <referredClinic>Eastside Health Laboratories</referredClinic>

 <referredBy>Michael Matthews</referredBy>

 <ThyroidFunction CPT=”84443”>

 <testCode = “TSH”>

 <CPT>84443</CPT>

 <LOINC></LOINC>

 <description>Thyroid Stimulating Hormone</description>

 <value>2</value>

 <units>mIU/L</units>

 </testCode>

 <testCode = “FT4”>

 <CPT>84439</CPT>

 <LOINC></LOINC>

 <description>T4, Free</description>

 <value>17</value>

 <units>pmol/L</units>

 </testCode>

 <testCode = “T3F”>

 <CPT>84481</CPT>

 <LOINC></LOINC>

 <description>T3, Free</description>

 <value>4</value>

 <units>pmol/L</units>

 </testCode>

 </ThyroidFunction>

 </test>

 <test = “Laboratory”>

 <CPT>80412</CPT>

 <testName>Corticotropic Releasing Hormone Stimulation Panel</testName>

 <referredName>Baljit Gupta</referredName>

 <referredClinic>Eastside Health Laboratories</referredClinic>

 <referredBy>Michael Matthews</referredBy>

 <CRH CPT=”80412”>

 <testCode = “ACH”>

 <CPT>82024</CPT>

 <LOINC></LOINC>

 <description>Adrenocorticotropic hormone</description>

 <value>3</value>

 <units>pmol/L</units>

 </testCode>

 <testCode = “CTL”>

 <CPT>82533</CPT>

 <LOINC></LOINC>

 <description>Cortisol</description>

 <value>522</value>

 <units>nmol/L</units>

 </testCode>

 </CRH>

 </test>

 <test = “Laboratory”>

 <CPT>85999</CPT>

 <testName>CoagulationBattery</testName>

 <referredName>Baljit Gupta</referredName>

 <referredClinic>Eastside Health Laboratories</referredClinic>

 <referredBy>Michael Matthews</referredBy>

 <Coagulation CPT=”85999”>

 <testCode = “PT”>

 <CPT>85610</CPT>

 <LOINC></LOINC>

 <description>Prothrombin time</description>

 <value>11</value>

 <units>s</units>

 </testCode>

 <testCode = “INR”>

 <CPT></CPT>

 <LOINC></LOINC>

 <description>INR</description>

 <value>1.0</value>

 <units></units>

 </testCode>

 <testCode = “Bleeding Time”>

 <CPT>85002</CPT>

 <LOINC></LOINC>

 <description>Bleeding time</description>

 <value>6</value>

 <units>min</units>

 </testCode>

 <testCode = “FIB”>

 <CPT>85384</CPT>

 <LOINC></LOINC>

 <description>Fibrinogen</description>

 <value>3</value>

 <units>g/L</units>

 </testCode>

 </Coagulation>

 </test>

Add b12 0.64nmol/L

Folate 2.1 ug/L

RPR – Non reactive

</encounter>

<Encounter>

 <dateOfEncounter>2009-01-15</dateOfEncounter>

 <physician>Michael Matthews</physician>

 <medicalBaseline>false</medicalBaseline>

 <medicalTest>false</medicalTest>

 <reportedSymptoms>Confusion, Becomes lost</reportedSymptoms>

 <workingDiagnosis>

 <preliminaryDiagnosis>

 <diagnosisDescription>Dementia</diagnosisDescription>

 <ICD9>331</ICD9>

 <ICD9Description> Other cerebral degenerations</ICD9Description>

 </preliminaryDiagnosis>

 <differentialDiagnosis>

 </differentialDiagnosis>

 </workingDiagnosis>

 <test = “ICD9 Procedure”>

 <ICD9 Procedure>94.02</ICD9 Procedure>

 <testName>Administration of psychologic test</testName>

 <referredName>Keil Fender</referredName>

 <referredClinic>UNMC</referredClinic>

 <referredBy>Michael Matthews</referredBy>

 <result = “Wechsler Memory Scale”>

 <description></description>

 <value></value>

 <units></units>

 </result>

 </test>

 <test = “ICD9 Procedure”>

 <ICD9 Procedure>88.91</ICD9 Procedure>

 <testName>Magnetic resonance imaging of brain and brain stem</testName> (Included here will be the **written reports of Radiologist**

 **To indicate likelihood of results to coincide with**

 **Alzheimer’s disease or dementia** – also included will be the ruling out of other potential causes of dementia-

 Type signs and symptoms which is an important step in making the diagnosis of alzheimers – to rule out other disease! It is difficult to describe the parameters of what a radiological definition of disease may be, this is beyond our scope and scans are used in accordance with all of the other prognostic indicators to arrive at a dioagnosis

MRI is nonspecific, although serial MRIs separated by >3months may show increased loss in medial temporal lobe (especially the Hippocampus – again depends on neuroradiologist opinion on sequential scans - think we should just place several scans and a brief opinion stating size differential that is suggestive of dementia

 <referredName>Sarah Yang</referredName>

 <referredClinic>Midtown Hospital</referredClinic>

 <referredBy>Michael Matthews</referredBy>

 <result = “DICOM 3.0 Image Data”

 ...

 </result>

 </test>

 <test = “Laboratory”>

 <CPT>85025</CPT>

 <testName>Blood count; complete (CBC), automated (Hgb, Hct, RBC, WBC and platelet count) and automated differential WBC count</testName>

 <referredName>Baljit Gupta</referredName>

 <referredClinic>Eastside Health Laboratories</referredClinic>

 <referredBy>Michael Matthews</referredBy>

 <CompleteBloodCount CPT=”85025”>

 ...

 </CompleteBloodCount>

 </test>

</Encounter>

<Encounter>

 <dateOfEncounter>2009-01-20</dateOfEncounter>

 <physician>Keil Fender</physician>

 <medicalBaseline>false</medicalBaseline>

 <reportedSymptoms>false</reportedSymptoms>

 <workingDiagnosis>

 <preliminaryDiagnosis>

 <diagnosisDescription>Dementia</diagnosisDescription>

 <ICD9>331</ICD9>

 <ICD9Description> Other cerebral degenerations</ICD9Description>

 </preliminaryDiagnosis>

 <differentialDiagnosis>

 <ICD9>331.0</ICD9>

 <ICD9Description>Alzheimer's disease</ICD9Description>

 </differentialDiagnosis>

 <clinicalDiagnosis>

 </clinicalDiagnosis>

 </workingDiagnosis>

 <test = “Cognative”>

 <CPT>97532</CPT>

 <testName>Alzheimer's Disease Assessment Scale-cognitive subscale</testName>

 <referredName>Mary Lubker</referredName>

 <referredClinic>Neurology Specialists</referredClinic>

 <referredBy>Michael Matthews</referredBy>

 <result>

 <testScore>???</testScore>

 <testNormalRange>????</testNormalRange>

 <testResult>abnormal</testResult>

 </result>

 </test>

 <test = “Genotyping SNPs”>

 <CPT> 83520(3), 83891(1), 83892(4), 83896(10), 83908(4), 83912(1)</CPT>

 <testName>ADmark Alzheimer's Evaluation</testName>

 <referredName> Mary Lubker </referredName>

 <referredClinic> Neurology Specialists </referredClinic>

 <referredBy>Michael Matthews</referredBy>

 <result> Pulling in this genetic data is excellent!

 <value>abnormal</value>

 <rs\_Number> rs429358</rs\_Number>

 <variant\_Synonyms>APOE4, NG\_007084.2:g.7903T>C</variant\_Synonyms>

 <gene\_Symbol>APOE</gene\_Symbol>

 <gene\_Name> apolipoprotein E</gene\_Name>

 <ENTREZ\_Gene\_ID>348</ENTREZ\_Gene\_ID>

 <Accession\_ID> NG\_007084.2</Accession\_ID>

 <Gene\_Region>coding\_region</Gene\_Region>

 <DNA\_Change>g.7903T>C</DNA\_Change>

 <DNA\_Change\_Type>Substitution</DNA\_Change\_Type>

 <Amino\_Acid\_Change>C112R</Amino\_Acid\_Change>

 <Amino\_Acid\_Change\_Type>Missense</Amino\_Acid\_Change\_Type>

 </result>

 </test>

</Encounter> Need to find a way to pull in biormarker associations – article I sent to you the other day… where can we pull these in from?

<Encounter>

 <dateOfEncounter>2009-01-30</dateOfEncounter>

 <physician>Kerri Fallen</physician>

 <medicalBaseline>false</medicalBaseline>

 <medicalTest>

 <test>

 <testName>MRI Brain Image</testName>

 <hippocampalVolume>???</hippocampalVolume>

 <hippocampalVolumeNormalRange>????</hippocampalVolumeNormalRange>

 <hippocampalVolumeTestResult>abnormal</hippocampalVolumeTestResult>

 <amyloidDeposit>???</amyloidDeposit>

 <amyloidDepositNormalRange>???</amyloidDepositNormalRange>

 <amyloidDepositResult>abnormal</amyloidDepositResult>

 </test> (These are again at the discretion of the Neuroradiologist and If we get involved with specific values we will be far out of our depth) If we really need them, I can search and find them, otherwise, take the radiologist’s word for either “changes suggestive”, or “progression” this information and anatomical association changes daily depending on association and specialist opinion – and will likely need consensus radiologist opinion to determine sensitivity/specificity. There is no “gold standard” other than an actual pathologic tissue sample that is diagnostic, just suggestive opinion

 </medicalTest>

 <reportedSymptoms>false</reportedSymptoms>

 <workingDiagnosis>Alzheimers Disease</workingDiagnosis>

</Encounter>

The basics we need to include somewhere :

1)Psychometric tests

2)Histology – unlikely to have any until death – the gold standard for diagnosis

3)Imaging : sequential “suggestive” changes are probably the best we can get

4)Blood flow imaging: hypoperfusion of the temporoparietal areas

And more anteriorly in frontotemporal dementia

5)Genetic Associations : we have these

Need to add :

Mosaic downs syndrome caused by mitotic nondisjunction in embryonic development

Need to include pharmacogenomic section

P450 isoenzyme ranges

6)CSF analysis : b amyloid decreased ; tau protein increased (both of these are the basics – don’t get too complicated)

7)Presentation and progression from encounter to encounter

8) Blood tests from encounter to encounter – in this case most are normal and this excludes other diseases as the cause for the condition – ultimately alzheimer’s disease is the diagnosis of exclusion

9)Predictive medicine :

Need to have genetic association data : data predicting drug efficacy and toxicity if available (no drugs in this example so not an issue)

Need to place biomarker associations and identify and pull from relevant data sources

Going to also need some text

Here is an example illustrating what I mean

Taken from :

<http://www.mtsamples.com/site/pages/sample.asp?Type=95-Radiology&Sample=566-Alzheimer+Disease>

Neuropsychological evaluation, 2/6/07, revealed: 1) well preserved intellectual functioning and orientation, 2) significant deficits in verbal and visual memory, proper naming, category fluency and working memory, 3)performances which were below expectations on tests of speed of reading, visual scanning, visual construction and clock drawing, 4)limited insight into the scope and magnitude of cognitive dysfunction. The findings indicated multiple areas of cerebral dysfunction. With the exception of the patient's report of minimal occupational dysfunction ( which may reflect poor insight), the clinical picture is consistent with a progressive dementia syndrome such as Alzheimer's disease. MRI brain 2/3/04, showed mild generalized atrophy, more severe in the occipital-parietal regions.

On 4/5/08, his performance on repeat neuropsychological evaluation was relatively stable. His verbal learning and delayed recognition were within normal limits, whereas delayed recall was "moderately severely" impaired. Immediate and delayed visual memory were slightly below expectations. Temporal orientation and expressive language skills were below expectation, especially in word retrieval. These findings were suggestive of particular, but not exclusive, involvement of the temporal lobes.

On 9/30/08, he was evaluated for a 5 minute spell of visual loss, OU. The episode occurred on Friday, 9/27/08, in the morning while sitting at his desk doing paperwork. He suddenly felt that his gaze was pulled toward a pile of letters; then a "curtain" came down over both visual fields, like "everything was in the shade." During the episode he felt fully alert and aware of his surroundings. He concurrently heard a "grating sound" in his head. After the episode, he made several phone calls, during which he reportedly sounded confused, and perseverated about opening a bank account. He then drove to visit his sister in Muscatine, Iowa, without accident. He was reportedly "normal" when he reached her house. He was able to perform Mass over the weekend without any difficulty. Neurologic examination, 9/30/08, was notable for: 1)category fluency score of 18items/60 sec. 2)VFFTC and EOM were intact. There was no RAPD, INO, loss of visual acuity. Glucose 178 (elevated), ESR ,Lipid profile, GS, CBC with differential, Carotid duplex scan, EKG, and EEG were all normal. MRI brain, 9/30/96, was unchanged from previous.

On 1/3/97, he had a 30 second spell of lightheadedness without vertigo, but with balance difficulty, after picking up a box of books. The episode was felt due to orthostatic changes.

1/8/97 neuropsychological evaluation was stable and his MMSE score was 25/30 (with deficits in visual construction, orientation, and 2/3 recall at 1 minute). Category fluency score 23 items/60 sec. Neurologic exam was notable for graphesthesia in the left hand.

In 2/97, he had episodes of anxiety, marked fluctuations in job performance and resigned his pastoral position. His neurologic exam was unchanged. An FDG-PET scan on 2/14/09 revealed decreased uptake in the right posterior temporal-parietal and lateral occipital regions.