Fall From Grace

### Prompt:  
  
"I have a 32-year-old previously healthy male patient admitted after sustaining burn injuries to his arm due to a fire caused by a model rocket mishap. Upon assessment, he displays unusual sensory changes, specifically complaining of an odd odor perception, stating that burned flesh smells like liquorice. His past medical history includes a self-admitted heroin overdose and a history of homelessness and nutritional neglect. He appears to have lost a significant amount of weight during his time without stable housing and has been on a restricted diet since his admission.   
  
Physical examination reveals multiple scars consistent with previous burns and possible injuries inflicted during childhood. He exhibits episodes of severe abdominal pain and rectal bleeding, which he claims began recently. His neurological examination is notable for cerebellar ataxia, indicated by poor coordination and difficulty judging distances. Initial investigations rule out common infections and drug-related toxicity.   
  
Given these clinical features and his significant reliance on a vegetarian diet since admission, what differential diagnoses should be considered? What specific tests would you recommend to confirm or exclude the most likely condition?"  
  
### Expected LLM Response:  
  
"In this case, the patient presents with a combination of sensory alterations, gastrointestinal symptoms, cerebellar ataxia, and significant burn injuries. The intriguing odor perception, rectal bleeding, and neurological signs point towards a metabolic or genetic disorder rather than a straightforward case of substance overdose or nutritional deficiency.   
  
Differential Diagnoses:  
  
1. \*\*Adult Refsum Disease\*\*: This condition is caused by defective phytanic acid metabolism leading to accumulation of phytanic acid; patients can present with neurological symptoms, including cerebellar ataxia, sensory disturbances, and dermatological issues. The patient's restricted diet, which may have high levels of chlorophyll from vegetables, could lead to increased levels of phytanic acid.  
  
2. \*\*Hereditary Sensory and Autonomic Neuropathy (HSAN)\*\*: This genetic disorder results in loss of sensation and problems with autonomic functions. While the patient's past heroin use might complicate the clinical picture, the bizarre sensory symptoms combined with the neurological examination raise suspicion for this diagnosis.  
  
3. \*\*Vitamin Overdose\*\*: Particularly vitamin A or other fat-soluble vitamins, from the high consumption of vegetables if such supplements are being taken. Symptoms might include visual disturbances and could explain some neurological symptoms.  
  
4. \*\*Acute Intermittent Porphyria\*\*: Although less likely given the presenting symptoms, porphyrias can lead to abdominal pain and neurological manifestations related to dietary factors.  
  
Recommended Diagnostic Tests:  
  
- \*\*Plasma Phytanic Acid Levels\*\*: This would confirm or rule out Adult Refsum Disease.  
- \*\*Genetic Testing\*\*: To look for mutations associated with Adult Refsum or other inherited metabolic disorders.  
- \*\*MRI of the Brain\*\*: To assess for any structural abnormalities associated with his neurological deficits.  
- \*\*Comprehensive Metabolic Panel\*\*: To check other vitamin levels and liver function.  
- \*\*Stool Testing\*\*: To assess for malabsorption or fat malabsorption, particularly in the context of potential vitamin overloads.  
  
Given the patient's complex presentation and dietary history, Adult Refsum Disease appears to be a leading suspect and warrants further investigation due to its potentially treatable nature."  
  
### DISEASE:  
Adult Refsum Disease