50 year old male

PC

- Difficulty with balance

HPC

- 18 month history of difficulty walking
- Falling over an d increasingly clumsy, bumping into things
- Now requiring two walking sticks, previously walking independently
- Family members comment that she is increasingly 'sounding drunk'
- No fevers, headaches or systemic complaints

PMHx

- Type 2 diabetes mellitus

PSHx

- Nil

THx

- Metformin 500mg BD

SHx

- Ex – EtOH, moderate amount and none for years
- Ex-Smoker 20 pack year history

FHx

- No known neurological disorder in parents, Father RIP myocardial infarction aged 53
- One of 4 siblings, older sister has history of neurological disorder affecting walking but no clear diagnosis

Examination

BP 140/70
HR 60
I+II+O

Sats 98% o/a
RR 14
Afebrile

Gait is unsteady, unable to tandem walk

Cranial Nerves

	R	L
I	N	N
II	N	N
III, IV, VI	N	N
	Nystagmus	Nystagmus
V	N	N
VII	N	N
VIII	N	N
IX,X,XI	N	N
XII	N	N

	R	L
Finger-nose	Impaired	Impaired
Heel-shin	Impaired	Impaired

Upper Limb

	R	L
Tone	N	N
Sh Ab	5	5
EF	5	5
EE	5	5
WE	5	5
FE	5	5
FDI	5	5
APB	5	5
Biceps	++	++
Triceps	++	++
Supinator	++	++

Lower Limb

	R	L
Tone	N	N
HF	5	5
HE	5	5
KF	5	5
KE	5	5
APF	5	5
ADF	5	5
AI	5	5
Knee	+++	+++
Ankle	+++	+++
Plantar	↔	↔

Rombergs

Pin Prick	*N*	*N*	*Unsteady with and*
JPS	*N*	*N*	*without eyes close*
Vib	*N*	*N*	

** Gaze evoked nystagmus, saccadic intrusion of smooth pursuit.*

Stop and think

1. **What are the potential causes for a cerebellar syndrome and how can they be categorised?**
2. **What are the first line investigations you would request?**
3. **How would you approach investigation of suspected genetic causes?**

Investigations

Bloods

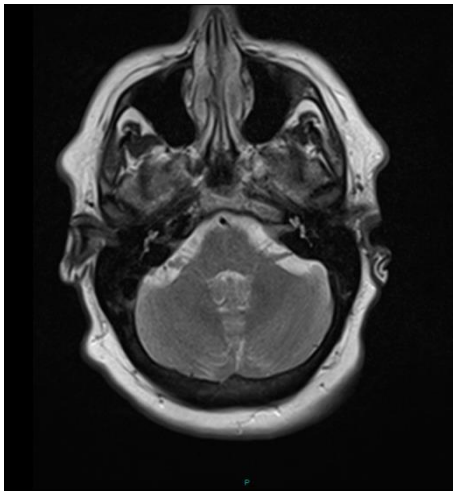
Hb	125	g/L
MCV	87	fL
WC	6	10*9/L
Plt	145	10*9/L
B12	220	ng/mL
Folate	16	ng/mL

Na	134	mmol/L
K	4.6	mmol/L
Ur	6.8	mmol/L
Creat	90	umol/L
CRP	3	mg/L

ALT	34	u/L
Alk P	50	u/L
Bili	18	umol/L
Alb	40	g/L
INR	1.0	

Paraneoplastic Anti-Hu	-ve
Vitamin E	N
Thiamine	N
Electrophoresis	N

Radiology



Normal appearances of cerebrum and cerebellum. No space occupying lesions.

Other

CT chest-abdomen-pelvis

No identified mass nor any significant lymphadenopathy.

Haematology	**Units**	**Normal Range**
Hb	g/L	120 – 160
MCV	fL	76-96
WC	10*9/L	4 – 11
PLT	10*9/L	140 – 450
B12	ng/mL	200 – 900
Folate	ng/mL	2-20
Renal Profile		
Na	mmol/L	133 – 146
K	mmol/L	3.5 – 5.3
Urea	mmol/L	2.5 – 7.8
Creatinine	umol/L	0 – 110
Liver Function		
ALT	u/L	11 – 55
Alk Phosphatase	u/L	30 – 130
Bilirubin	umol/L	0 – 21
Albumin	g/L	35 – 50
INR		1.0
Bone Profile		
Calcium	mmol/L	2.18 – 2.62
Phosphate	mmol/L	0.8 – 1.5
Other		
CK	u/L	25 - 200
Mg2+	mmol/l	0.75 – 1.0
Cortisol	nmol/l	130 - 690
T4	mmol/L	70-140
TSH	mu/L	0.5 - 5
C-Reactive Protein	mg/L	0 - 10
ESR	mm in 1 hr	3 - 9
Blood gas		
pH		7.35 – 7.45
PaO2	>10.6	kPa
PaCO2	4.7 - 6	kPa
CSF		
Opening pressure	cmH20	14 – 20
WC		<5
RC		<5
Protein	g/L	0.2 – 0.4
Glucose	Mmol	60 – 100% serum
Serum glucose	Mmol/L	4 – 7.8