Cerebellar Ataxia in Children and Adults:

A Vestibular Physician's Deep Review of Mechanism, Diagnosis, and Management

Vestibular Medicine for Vestibular Physicians

Central Vestibular Pathology — Module 3.3

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How to Use This Review

This literature review forms part of the Vestibular Medicine for Vestibular Physicians series published by the Australian Dizziness Clinics Education Hub. It is written for vestibular physicians, neuro-otologists, advanced ENT trainees, and vestibular physiotherapists working at the deep end of central vestibular practice, where a working command of mechanism, classification, and atypical presentations is expected rather than optional.

The review is dense by design — intended as a 30–40 minute deep read or a desktop reference. It is supported by an A4 clinician cheat sheet, short-form clinician videos, audio episodes, and a patient information leaflet within the same Education Hub module.

Callout Box Guide

□ **Key Point:** Foundational concepts and summary statements that anchor the core clinical content of each section.

□ **Clinical Insight:** Clinically relevant observations for direct application in assessment and management.

□ **Clinical Pearl:** High-yield memorable clinical points — the take-home messages most likely to change practice.

□ **Important:** Red flags, atypical presentations, and critical safety points requiring escalation or imaging.

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I. Introduction and Epidemiology

Cerebellar ataxia is a clinical syndrome of incoordination arising from disease of the cerebellum or its afferent and efferent connections, rather than a single disease entity [1,3]. The term ataxia derives from the Greek a-taxis ('lack of order') and has been used since antiquity, but the modern conception dates to the nineteenth century: Friedreich's 1863 description of an inherited early-onset ataxia with spinal-cord degeneration, and Pierre Marie's 1893 delineation of a dominantly inherited cerebellar ataxia distinct from it [1,2]. Through the twentieth century the field separated hereditary, sporadic-degenerative, acquired and, more recently, immune-mediated causes — a four-way partition that still structures the diagnostic approach today [3,5,13,34].

For the vestibular physician the importance of cerebellar ataxia is disproportionate to its frequency. In a balance clinic it represents a small minority of presentations relative to peripheral vestibular disease, but it is the diagnosis that is most often missed, most often mistaken for a peripheral disorder, and most often time-critical [27,32]. An acute cerebellar stroke can present as an isolated acute vestibular syndrome that mimics vestibular neuritis, and the bedside discrimination is life-saving [27,32].

Cerebellar ataxias are individually rare but collectively a significant cause of neurological disability [7]. Prevalence varies markedly by aetiology and geography. The combined prevalence of hereditary ataxias is of the order of a few per 100,000: autosomal dominant spinocerebellar ataxias (SCAs) are estimated at roughly 1–5 per 100,000 worldwide, with European pooled estimates near 2.7 per 100,000 [5,7]. Friedreich ataxia (FRDA), the commonest autosomal recessive ataxia, affects approximately 1 in 30,000 people of European ancestry [9]. Founder effects shape the regional picture — SCA3 (Machado–Joseph disease) is the single most prevalent dominant ataxia globally [10], while SCA2 clusters in parts of India and in Cuba, where one of the largest founder populations in the world is concentrated [8,11].

Among the sporadic degenerative ataxias, multiple system atrophy of cerebellar type (MSA-C) is the key differential, with an overall MSA prevalence near 2–5 per 100,000 [12]. Idiopathic late-onset cerebellar ataxia (ILOCA) remains a diagnosis of exclusion; on longitudinal follow-up a substantial fraction of patients initially labelled ILOCA are reclassified as MSA or as a defined genetic ataxia [13,14]. The acquired ataxias — vascular, neoplastic, toxic, nutritional and immune — are heterogeneous and not systematically enumerated, but two entities deserve emphasis. Gluten ataxia has been reported among a notable proportion of otherwise idiopathic sporadic ataxias [15,16], and CANVAS (cerebellar ataxia, neuropathy and vestibular areflexia syndrome), caused by biallelic RFC1 repeat expansion, is now recognised as a common cause of late-onset ataxia and a particularly important one for vestibular practice given its bilateral vestibular failure [17,18].

In children the epidemiology is dominated by transient, post-infectious disease. A systematic review estimated the prevalence of all-cause childhood ataxia at approximately 26 per 100,000 [19]. Many paediatric presentations — acute post-infectious cerebellitis being the prototype — are self-limiting and recover fully, whereas the remainder comprise rare genetic and metabolic disorders, several of which are treatable and must not be missed [19,20].

□ **Key Point:** Cerebellar ataxia is a syndrome, not a diagnosis. The clinical task is always two-step: confirm that the incoordination is cerebellar, then determine the cause along the hereditary / sporadic-degenerative / acquired / immune axis [3,13,34].

II. Pathophysiology — Cerebellar Circuitry and the Mechanisms of Ataxia

The cerebellum coordinates movement by comparing intended motor commands with sensory feedback — vestibular, proprioceptive and visual — and issuing corrective, predictive signals. Anatomically it is a three-lobed structure (anterior, posterior and flocculonodular lobes) folded into folia and divided by the primary and posterolateral fissures, as shown in Figure 1.

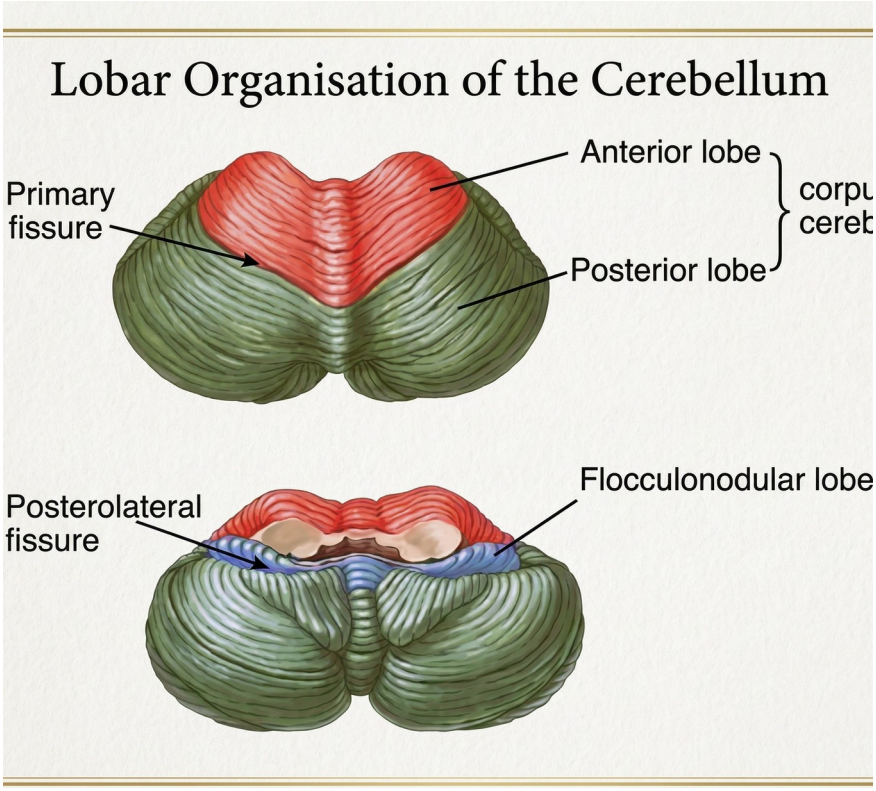

Figure 1. External (lobar) organisation of the cerebellum — superior (top) and inferior (bottom) views.
Source: OpenStax, Anatomy & Physiology (Rice University). Licensed under CC BY 4.0.

Functionally the cerebellum is organised into three zones whose involvement predicts the clinical syndrome: the vestibulocerebellum (flocculonodular lobe) for balance and eye movements; the spinocerebellum (vermis and paravermal hemispheres) for gait and axial coordination; and the cerebrocerebellum (lateral hemispheres) for skilled limb movement, motor planning and cognition [21,22]. Vermis lesions produce truncal and gait ataxia; hemispheric lesions produce appendicular ataxia, dysarthria and intention tremor; flocculonodular lesions produce nystagmus, impaired smooth pursuit and vertigo (Figure 2) [22,23].

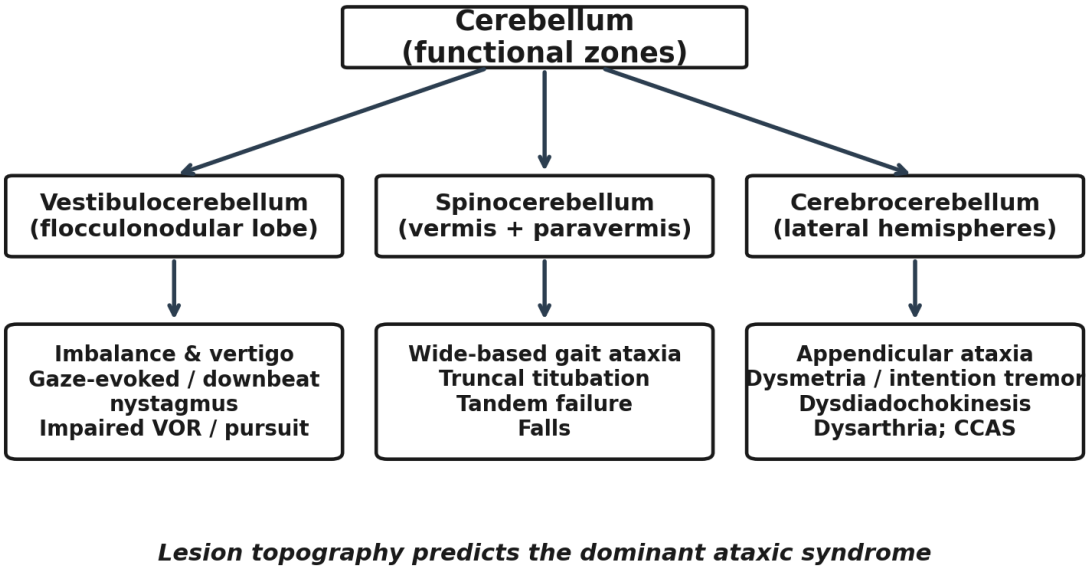

Figure 2. Functional zones of the cerebellum mapped to their dominant ataxic signs.

Source: Adapted from Stoodley and Schmahmann [22] and Manto [21].

At the cellular level the Purkinje cell is the pivotal output neuron, projecting inhibitory signals to the deep cerebellar nuclei, which in turn drive the motor pathways (Figure 3). The overwhelming majority of cerebellar ataxias converge on dysfunction or loss of Purkinje cells, or disruption of their inputs and outputs; the result is a failure to modulate motor commands and the disordered movement that defines ataxia [3,23].

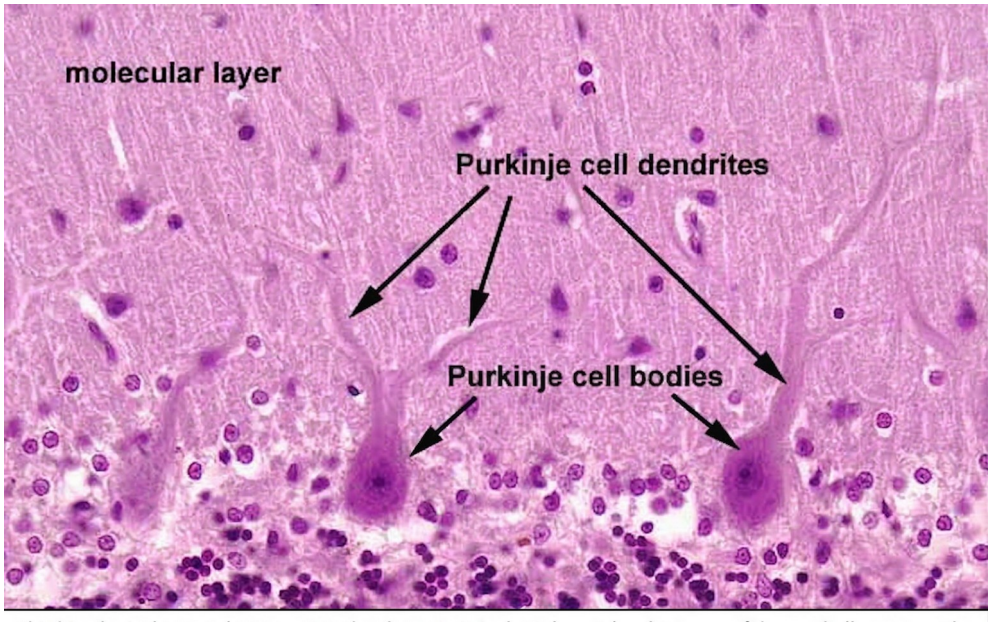

Figure 3. Histology of the cerebellar cortex — the Purkinje-cell layer between the molecular and granular layers.
Source: Wikimedia Commons — cerebellar cortex, haematoxylin–eosin stained section.

Hereditary degenerative ataxias are predominantly toxic gain-of-function disorders. The polyglutamine SCAs (SCA1, 2, 3, 6, 7 and others) arise from CAG-repeat expansions that yield misfolded ataxins which aggregate in Purkinje and brainstem neurons, disrupting transcription and proteostasis and producing progressive atrophy [5,6,24]. Friedreich ataxia is mechanistically distinct: a GAA intronic expansion in FXN silences frataxin, causing mitochondrial iron accumulation, oxidative stress and degeneration centred on the dorsal root ganglia, dorsal columns, spinocerebellar tracts and dentate nucleus [25,26]. The concept of cerebellar reserve — the capacity of surviving circuitry to compensate until a threshold of loss is crossed — explains both the delayed clinical onset of slowly progressive disease and the therapeutic urgency in potentially reversible causes [30]. These mechanisms, together with the immune and structural routes below, converge on a single final common pathway (Figure 4).

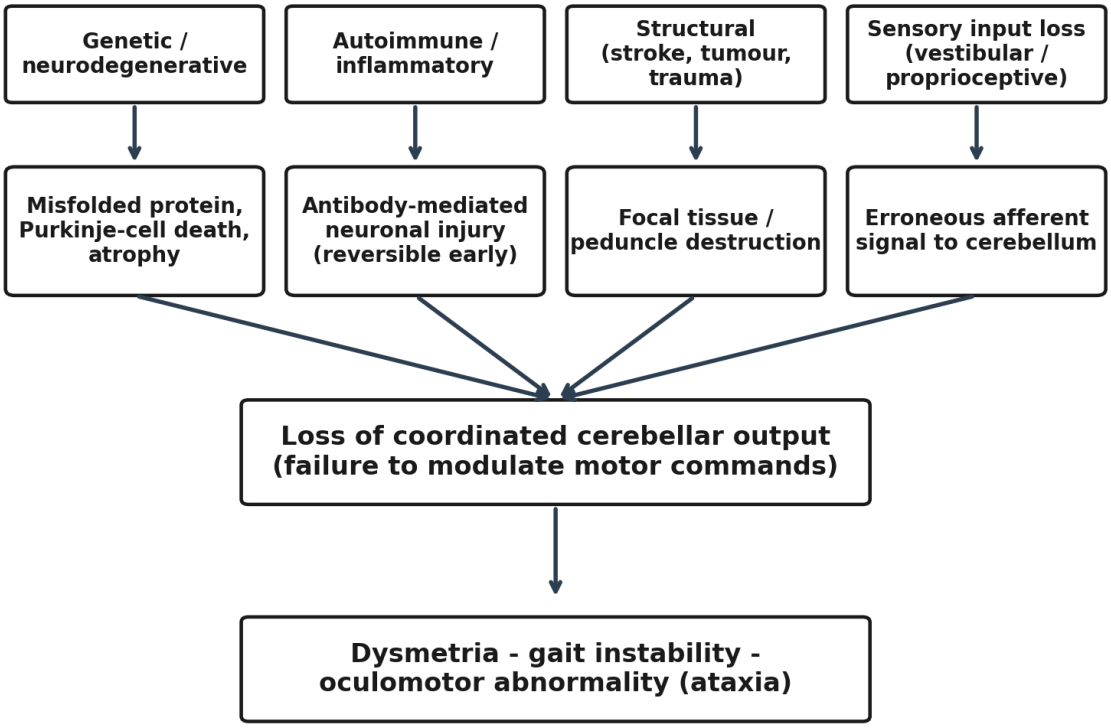

Figure 4. Pathophysiological routes to ataxia converging on a final common pathway.
Source: Adapted from Manto and Marmolino [3] and Mitoma et al. [28].

Acquired structural lesions damage the cerebellum focally. Infarction or haemorrhage in a cerebellar arterial territory — supplied by the posterior inferior, anterior inferior and superior cerebellar arteries of the vertebrobasilar circulation (Figure 5) — produces ipsilateral limb ataxia, gait ataxia, dysarthria or vestibulocerebellar signs according to location; large acute lesions cause immediate severe ataxia, and posterior-fossa haemorrhage can be rapidly fatal through brainstem compression and obstructive hydrocephalus [27].

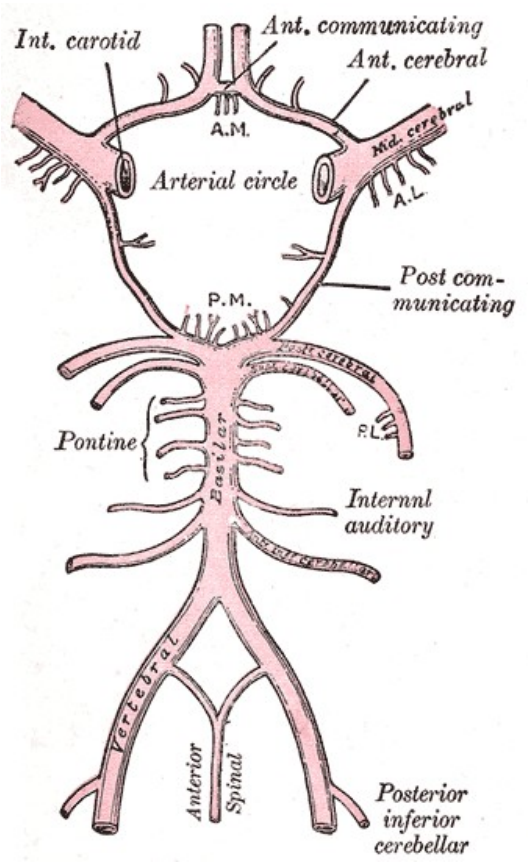

Figure 5. Posterior (vertebrobasilar) circulation supplying the cerebellum and brainstem.
Source: Gray's Anatomy of the Human Body (public domain).

Immune-mediated ataxias result from antibody attack on cerebellar neurons: paraneoplastic cerebellar degeneration (anti-Yo, anti-Hu, anti-CV2) destroys Purkinje cells rapidly, whereas non-paraneoplastic forms associated with anti-GAD or transglutaminase-6 antibodies may be at least partly reversible if treated before irreversible cell loss occurs [28,29,30]. Finally, lesions of the vestibulocerebellum or of vestibular afferents generate a syndrome dominated by imbalance, vertigo and nystagmus rather than limb ataxia. Downbeat nystagmus reflects flocculus dysfunction, and in CANVAS the combination of bilateral vestibular failure and cerebellar degeneration produces a profound, vision-dependent ataxia that worsens dramatically in darkness or on uneven ground [17,18].

□ **Clinical Insight:** Whatever the upstream cause — genetic, immune, structural or sensory — the final common pathway is loss of coordinated cerebellar output. This is why heterogeneous diseases share a stereotyped examination, and why topography (which zone) often localises better than aetiology at the bedside [3,22].

III. Clinical Features and Onset Patterns

Cerebellar ataxia presents as a constellation of motor-coordination deficits. Patients report imbalance and a lurching, 'drunken' gait, clumsiness of the hands, slurred speech, deteriorating handwriting and frequent falls; oscillopsia and diplopia occur when the vestibulocerebellum or ocular-motor control is involved [3,23]. The cardinal examination signs are gait ataxia (wide-based, irregular stride, impaired or impossible tandem walking, truncal titubation); appendicular ataxia (dysmetria on finger–nose and heel–knee–shin testing, dysdiadochokinesis, decomposition of movement); intention tremor; scanning dysarthria; gaze-evoked or downbeat nystagmus; and, less consistently, hypotonia with pendular reflexes [22,23].

The cerebellum is not purely motor. Schmahmann's cerebellar cognitive affective syndrome (CCAS) — executive dysfunction, impaired visuospatial cognition, language disturbance and affective change ranging from blunting to disinhibition — reflects disruption of cerebellar connections with prefrontal and

limbic cortex and should be screened for, including with the dedicated CCAS scale, in any patient with cerebellar disease [23,31].

The distribution of signs follows topography. Vermis lesions yield prominent truncal and gait ataxia with relatively preserved limb coordination; hemispheric lesions yield limb ataxia and intention tremor; pure vestibulocerebellar dysfunction yields vertigo, gait ataxia and nystagmus with little appendicular involvement [22,23]. Because many ataxias are multisystem disorders, associated features are diagnostically valuable: peripheral neuropathy (FRDA, CANVAS, vitamin-E deficiency), pyramidal signs (FRDA classically combines extensor plantar responses with absent ankle jerks; some SCAs), extrapyramidal features and slow saccades (SCA2, SCA3), autonomic failure (pointing to MSA), and systemic clues such as cardiomyopathy and scoliosis in FRDA or telangiectasia and immunodeficiency in ataxia–telangiectasia [18,38,46,47].

Onset tempo is the single most useful first filter. Acute onset over hours to days suggests stroke, haemorrhage, intoxication, Wernicke encephalopathy or acute demyelination. Subacute onset over weeks to a few months points to immune, paraneoplastic, infectious or, occasionally, prion disease. Chronic insidious onset over months to years is the signature of hereditary and sporadic degenerative ataxias [34,35]. This temporal triage shapes the entire investigation strategy and is reproduced in Figure 6.

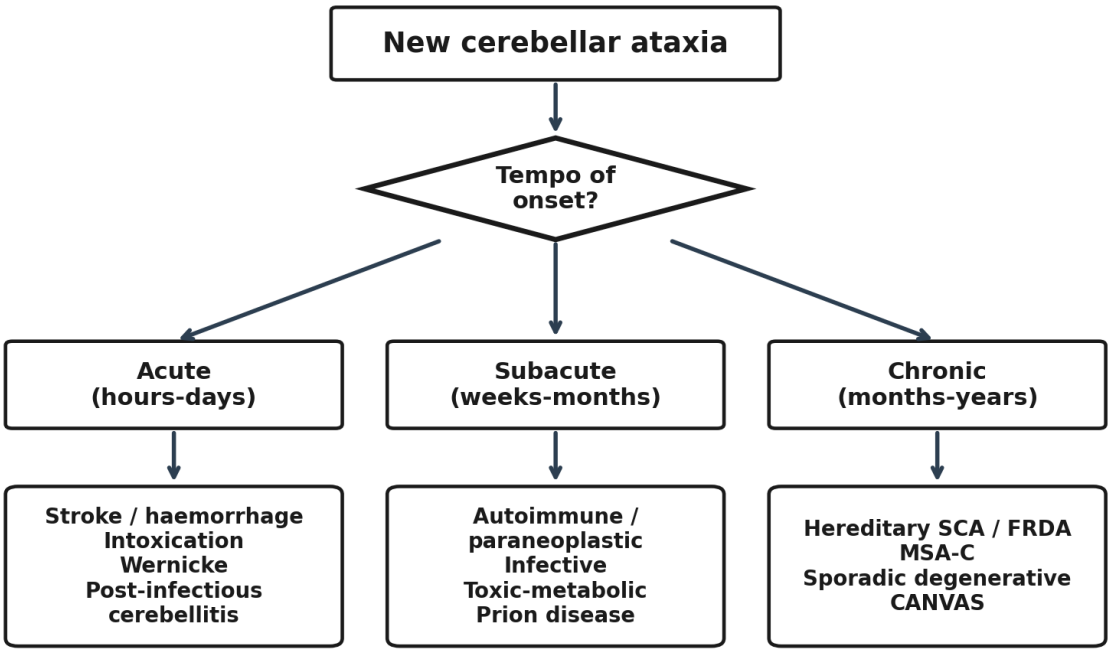

Figure 6. Onset tempo as the first diagnostic filter in cerebellar ataxia.
Source: Adapted from de Silva et al. [34] and Klockgether [13].

Children deserve separate emphasis. Acute childhood ataxia is usually benign and post-infectious, with full recovery the norm, but the differential includes ingestion, posterior-fossa tumour, opsoclonus–myoclonus and treatable metabolic disease, so a dramatic presentation must not be assumed to be benign without appropriate exclusion [19,20]. Standardised rating — the Scale for the Assessment and Rating of Ataxia (SARA) and the older International Cooperative Ataxia Rating Scale (ICARS) — should be recorded at baseline in both children and adults to quantify severity and track change [36,37].

□ **Important:** Acute cerebellar ataxia with vertigo is a posterior-circulation stroke until proven otherwise. A normal horizontal head-impulse test with direction-changing nystagmus and/or skew deviation (a central HINTS pattern) is more sensitive than early diffusion-weighted MRI and mandates urgent imaging and stroke pathways [27,32].

IV. Classification and Diagnostic Criteria

Because 'cerebellar ataxia' is a syndrome, there is no single diagnostic criterion; diagnosis proceeds by confirming cerebellar incoordination on examination — not explained by weakness or sensory loss — and then classifying by duration, family history and associated features [4,34]. Acquired causes are excluded first in any recent-onset case, because several are treatable and time-critical [34,35]. Where there is a positive family history or consanguinity, hereditary ataxia is pursued with targeted then broad genetic testing; where no cause emerges after comprehensive work-up, a sporadic degenerative ataxia is diagnosed by exclusion [13,35]. The genetic ataxias are conventionally grouped by inheritance, and the four tables below summarise the dominant, recessive, X-linked and mitochondrial groups that the vestibular physician should recognise.

Table 1. Autosomal dominant cerebellar ataxias (spinocerebellar ataxias and dominant episodic ataxias).

Autosomal dominant ataxias usually present in adulthood with progressive cerebellar dysfunction; most are trinucleotide-repeat SCAs, several with multisystem features [5,6,21].

Ataxia (gene)	Genetic / pathology	Typical age at onset	Chief clinical features	Characteristic features	Geographic / prevalence notes
SCA3 – Machado–Joseph (ATXN3)	CAG repeat (polyglutamine expansion)	Adult onset	Progressive gait and limb ataxia, dysarthria, often peripheral neuropathy and pyramidal signs	Most common AD ataxia worldwide; external ophthalmoplegia, dystonia or parkinsonism, cerebellar atrophy	Highest frequencies in certain founder populations (e.g. Portuguese/Azorean, Japanese, Brazilian); present worldwide
SCA2 (ATXN2)	CAG repeat (polyQ expansion)	Adult onset; slow progression over ~10–20 years	Gait ataxia, dysarthria, neuropathy with areflexia, mild cognitive decline	Markedly slow or absent saccadic eye movements; sleep disturbance and tremor can occur	Particularly common in some regions (e.g. parts of India and Cuba); also seen globally
SCA1 (ATXN1)	CAG repeat (polyQ expansion)	Typically 30s–40s	Progressive cerebellar ataxia, dysarthria, gait instability, pyramidal signs (hyperreflexia, extensor plantars)	Early bulbar dysfunction (dysphagia), strong pyramidal involvement; faster progression, wheelchair within ~3–15 years	Worldwide, no single founder region, but families described in many populations
SCA6 (CACNA1A)	CAG repeat (polyQ expansion) in calcium channel	Late adult onset (40s–50s)	“Pure” cerebellar ataxia – gait and limb ataxia with dysarthria; strength and reflexes usually normal	Pure cerebellar syndrome with pronounced gaze-evoked nystagmus, very slow progression; many remain ambulant >10 years	More frequent in some European and Japanese cohorts; recognised worldwide
SCA7 (ATXN7)	CAG repeat (polyQ expansion)	Young adulthood (earlier in large expansions)	Progressive ataxia plus visual impairment (blurred vision, field loss)	Cone–rod retinal dystrophy with early loss of visual acuity and colour vision; visual symptoms may precede ataxia; can lead to early blindness	Clusters reported in Scandinavia, South Africa (Afrikaner), and elsewhere due to founder effects
SCA8 (ATXN8OS/AT	CTG repeat expansion in	Adult onset	Cerebellar ataxia, dysarthria,	Reduced penetrance; some	Reported mainly in North

XN8)	non-coding region		dysphagia; often mild and very slowly progressive	expansion carriers asymptomatic. Mixed spastic–ataxic dysarthria, slight sensory loss; incomplete dominance	America and Europe; true pathogenicity debated in some cohorts
SCA10 (ATXN10)	ATTCT pentanucleotide repeat expansion	Adolescent or adult onset	Ataxia with dysarthria; many patients develop generalised epileptic seizures a few years after onset	Distinctive combination of cerebellar ataxia and generalised motor seizures; MRI with cerebellar atrophy, reflexes often normal	Strong founder effect in Latin American populations (especially Mexico, Brazil); rare elsewhere
SCA12 (PPP2R2B)	CAG repeat expansion (5′ UTR)	Adult onset (mean ~40 years)	Action tremor of upper limbs often precedes mild gait ataxia; later dysarthria and ataxia	Tremor–ataxia syndrome: early prominent action tremor resembling essential tremor; protein phosphatase gene involvement	Notably common in a North Indian founder population; rare elsewhere
SCA17 (TBP)	CAG/CAA repeat (polyQ expansion) in TBP	Young adulthood	Mixed cerebellar, extrapyramidal and cognitive syndrome: ataxia, seizures, dementia, psychiatric symptoms	“Huntington-like” ataxia with dementia, chorea or dystonia, psychosis and seizures; broader phenotype than pure SCAs	Families reported worldwide; many series from Europe and East Asia
DRPLA (ATN1)	CAG repeat (polyQ expansion)	Juvenile or young adult; anticipation common	Progressive ataxia with myoclonus, epilepsy, choreoathetosis and dementia	Dentatorubral–pallidoluysian atrophy; striking anticipation with earlier, more severe disease in successive generations	Predominantly East Asia (especially Japan), rare in other populations
Episodic Ataxia type 2 (EA2, CACNA1A)	Various CACNA1A mutations (calcium channel)	Usually childhood/teen years	Recurrent episodic ataxia (hours), often triggered by stress/exertion; interictal nystagmus or mild ataxia	Acetazolamide-responsive in many cases; interictal gaze-evoked nystagmus; allelic to SCA6 but episodic rather than steadily progressive	Cases reported worldwide; often familial AD
Episodic Ataxia type 1 (EA1, KCNA1)	Point mutations in KCNA1 (K^+ channel)	Childhood onset	Brief episodes of ataxia (seconds–minutes) often with startle or exercise; baseline myokymia	Short-duration attacks; patients neurologically normal between attacks apart from neuromyotonia. Responsive to acetazolamide or anticonvulsants in many	Rare; scattered families worldwide, originally described in North America

Table 2. Autosomal recessive cerebellar ataxias.

Recessive ataxias more often begin in childhood or adolescence, frequently involve multiple systems, and include several metabolically treatable disorders that must be actively sought [20,47].

Ataxia (gene)	Genetic / pathology	Typical age at onset	Chief clinical features	Characteristic features	Geographic / prevalence notes
Friedreich ataxia (FXN)	GAA intronic repeat expansion →	Childhood or adolescenc	Progressive gait and limb ataxia, dysarthria, areflexia,	Most common hereditary ataxia (~½ of all hereditary	Prevalence ~2–4 per 100 000 in European-

	frataxin deficiency, mitochondrial dysfunction	e	dorsal column sensory loss, extensor plantar responses	cases). Combination of peripheral neuropathy (areflexia) and pyramidal signs (Babinski). Hypertrophic cardiomyopathy, diabetes, scoliosis and pes cavus frequent	ancestry populations; rare in East Asia and Africa
Ataxia–telangiectasia (ATM)	ATM mutation → DNA repair defect	Early childhood (toddler)	Progressive cerebellar ataxia and dysarthria, immune deficiency with recurrent sinopulmonary infections	Oculocutaneous telangiectasias (sclera/skin) by age 3–6; very high AFP, marked radiosensitivity and cancer risk (leukaemia/lymphoma)	Worldwide but rare; higher incidence in some consanguineous populations
Ataxia with oculomotor apraxia type 1 (AOA1, APTX)	Aprataxin mutation → DNA repair defect	Childhood (~age 4)	Progressive cerebellar ataxia, oculomotor apraxia (head thrusts to initiate gaze), distal weakness and areflexic neuropathy, foot deformities	Early chorea/myoclonus may occur then fade; low serum albumin and high cholesterol; wheelchair usually needed 10–15 years after onset	Reported in multiple regions with clusters (e.g. Portugal, Japan); rare overall
Ataxia with oculomotor apraxia type 2 (AOA2, SETX)	Senataxin mutation → DNA repair defect	Later childhood/teens (~10–20)	Progressive ataxia, oculomotor apraxia, peripheral neuropathy with areflexia and limb atrophy; chorea and head tremor can persist	Elevated AFP distinguishes AOA2 from AOA1; serum albumin usually normal, mild hypercholesterolaemia. Slower progression than AOA1	Europe and Japan most reported; rare worldwide
AR spastic ataxia of Charlevoix–Saguenay (ARSACS, SACS)	Sacsin mutation	Early childhood (toddler walking age)	Ataxia with leg spasticity, peripheral neuropathy, dysarthria, distal wasting; prominent pyramidal signs	Thick myelinated retinal nerve fibres encasing vessels (fundus); lifelong gait abnormality; frequent finger and foot deformities; often wheelchair-bound by mid-adulthood	Originally described in Quebec founder population; now recognised in Europe, North Africa and elsewhere
CANVAS (RFC1)	Biallelic AAGGG intronic repeat in RFC1	Late onset (>40 years)	Sensory neuropathy (worse in dark), bilateral vestibular areflexia and cerebellar ataxia with falls and oscillopsia	Classic triad: cerebellar ataxia + sensory neuropathy + vestibular loss; emerging as a common cause of late-onset AR ataxia	Reported particularly in UK, Europe and Australia; likely under-recognised
Wilson disease (ATP7B)	Copper transport defect → toxic copper deposition	Variable: childhood to young adult	Cerebellar ataxia and dysarthria often with dystonia, tremor, parkinsonism or psychiatric symptoms; hepatic disease common	Kayser–Fleischer rings, “wing-beating” tremor, dystonic facies. Treatable with chelators (penicillamine) and zinc; neurologic and hepatic features	Worldwide; higher frequency in some regions (e.g. Mediterranean, Middle East, parts of Asia)
Ataxia with vitamin E deficiency	α-Tocopherol transfer protein defect → very	Teens or early adulthood	Progressive ataxia with peripheral neuropathy (loss of	Extremely low serum vitamin E; lifelong high-dose vitamin E	Described in Mediterranean and Middle

(AVED, TTPA)	low vitamin E		reflexes and proprioception), resembling FRDA; dysarthria and gait instability	can stabilise/improve symptoms. May have polyneuropathy, extensor plantars, cardiomyopathy and sometimes retinal changes	Eastern populations; treatable and likely underdiagnosed
Abetalipoproteinemia (MTP)	Microsomal triglyceride transfer protein defect → fat and vitamin E malabsorption	Infancy (failure to thrive, steatorrhoea); neurological in childhood	Progressive ataxia, areflexia, peripheral neuropathy and retinal degeneration (night blindness) due to fat-soluble vitamin deficiency	Acanthocytosis on blood smear, very low cholesterol; neurologic damage largely due to vitamin E deficiency; high-dose vitamin E and fat modification can improve outcome	Very rare; most data from case series in consanguineous families
Refsum disease (PHYH or PEX7)	Peroxisomal phytanic acid oxidation defect	Teens or early adulthood	Peripheral neuropathy, cerebellar ataxia, retinitis pigmentosa; ichthyosis and hearing loss common	Elevated phytanic acid; dietary restriction of dairy/beef/fish halts progression; cardiac arrhythmias/heart block can occur if untreated	Uncommon; more reports in Northern Europe; treatable metabolic leukodystrophy
Cerebrotendinous xanthomatosis (CTX, CYP27A1)	Sterol 27-hydroxylase defect → bile acid and cholesterol metabolism abnormality	Neurologic usually adolescence/early adult; GI/cataracts in childhood	Progressive ataxia and cognitive decline; childhood chronic diarrhoea, juvenile cataracts; tendon xanthomas (Achilles)	Treatable leukodystrophy: chenodeoxycholic acid can halt progression. Ataxia, dementia, pyramidal signs, polyneuropathy	Worldwide but very rare; likely underdiagnosed; founder mutations in some populations
Coenzyme Q10 deficiency ataxia (e.g. ADCK3)	CoQ10 biosynthesis defect	Childhood or early adulthood (some infantile)	Cerebellar ataxia with exercise intolerance; may have seizures, myopathy or neuropathy; developmental delay in severe forms	Ubiquinone-responsive ataxia: oral CoQ10 can markedly improve symptoms; suspect in young ataxia with myopathy/seizures and low CoQ10 levels	Rare; scattered cases worldwide; important as treatable
Niemann–Pick disease type C (NPC1/2)	Lysosomal cholesterol trafficking defect	Juvenile (often 10–20 years)	Ataxia, dysarthria, cognitive decline; hepatosplenomegaly often precedes neuro signs; progressive dementia and dystonia	Vertical supranuclear gaze palsy and cataplexy are highly characteristic; miglustat may slow progression	Worldwide; many cases in consanguineous or founder populations

Table 3. X-linked cerebellar ataxias.

Ataxia (gene)	Genetic / pathology	Typical age at onset	Chief clinical features	Characteristic features	Geographic / prevalence notes
Fragile X–associated tremor/ataxia syndrome (FXTAS, FMR1)	FMR1 CGG premutation (55–200 repeats), X-linked	Late adult (>50 years); males >> females	Progressive intention tremor, gait ataxia, parkinsonism; cognitive decline (executive dysfunction), peripheral neuropathy	Most common X-linked adult ataxia. Premutation carriers; MRI with middle cerebellar peduncle white-matter lesions. Females affected more mildly	Seen worldwide wherever Fragile X premutation occurs; relatively frequent among older male premutation

					carriers
Rett syndrome (MECP2, X-dominant)	MECP2 loss-of-function (usually de novo)	6–18 months after normal early infancy (girls)	Neurodevelopmental regression, loss of speech, gait ataxia, apraxia, seizures, intellectual disability; hand stereotypies	Predominantly females (male lethal). Early deceleration of head growth (microcephaly), autism-like features and breathing abnormalities; ataxia within a complex neurodevelopmental picture	Global; prevalence around 1 in 10 000 girls
X-linked spinocerebellar ataxia 1 (SCAX1, ATP2B3)	ATP2B3 mutation (Xq28), X-recessive	Congenital or infantile	Non-progressive congenital cerebellar ataxia, newborn hypotonia, delayed milestones, nystagmus, speech delay; mild intellectual disability in some	Lifelong but non-progressive ataxia in boys; slow eye movements (smooth pursuit). Also called X-linked congenital ataxia	Extremely rare; a few families described
X-linked ataxia with spasticity (SCAX2)	Unknown X-linked mutation, X-recessive	Infancy	Ataxia with lower limb spasticity, head/neck tremor, motor delay and hypotonia; severe disability in childhood	Combination of cerebellar ataxia and pyramidal signs from birth; often intellectual disability and premature death	Very rare; only a small number of families
X-linked ataxia with deafness (SCAX3)	Unknown X-linked mutation, X-recessive	Infancy	Ataxia with hypotonia and congenital sensorineural deafness; delayed motor development, possible strabismus; progressive with early childhood mortality	Unique combination of bilateral deafness and cerebellar ataxia from infancy; severe hypotonia	Reported in a few pedigrees only
X-linked ataxia with late dementia (SCAX4)	Unknown X-linked mutation, X-recessive	Childhood (ataxia), dementia in 30s	Mild ataxia and tremor in childhood; later progressive fronto-subcortical dementia in mid-adulthood	Biphasic course: relatively static early ataxia then later dementia; premature death by ~60s	Extremely rare (single family described)
X-linked congenital ataxia (SCAX5)	Unknown X-linked mutation, X-recessive	Congenital	Cerebellar ataxia with hypotonia, nystagmus and dysarthria from infancy; some motor improvement with age	Non-progressive or improving course; ataxia present from birth but partially ameliorates in later childhood; gene not yet identified	Very rare, small number of reported families
X-linked sideroblastic anaemia with ataxia (SCAX6, ABCB7)	ABCB7 mutation, X-recessive	Infancy	Cerebellar ataxia plus microcytic sideroblastic anaemia (fatigue, pallor); may have nystagmus, dysarthria, mild spasticity	Unique combination of bone-marrow failure with ringed sideroblasts and early-onset ataxia; anaemia partly pyridoxine-responsive	Very rare; limited families described
Oligophrenin-1 syndrome (OPHN1)	OPHN1 mutation, X-recessive	Early childhood	Cerebellar ataxia, developmental delay, hypotonia, intellectual disability; seizures in some	Cerebellar vermis hypoplasia on MRI; X-linked intellectual disability with ataxia, hypotonia and	Rare; multiple families worldwide

				strabismus	
CASK-related syndrome (CASK, X-dominant, male-lethal)	CASK mutation (usually de novo)	Infancy (girls)	Infantile hypotonia, microcephaly, developmental delay; survivors develop cerebellar ataxia, profound intellectual disability and seizures	Microcephaly with pontocerebellar hypoplasia; severe neurodevelopmental syndrome (MICHA) in females; hemizygous males usually die in infancy	Sporadic de novo cases worldwide
Christianson syndrome (SLC9A6)	SLC9A6 mutation, X-recessive	Early childhood after initial normal infancy	Developmental regression, progressive ataxia, epilepsy, absent speech, cognitive impairment, hyperactivity and autism-like traits	"Angelman-like" phenotype: happy demeanour, hyperactivity, ataxia, seizures, ophthalmoplegia, microcephaly	Very rare; described mainly in small family series

Table 4. Mitochondrial cerebellar ataxias.

Mitochondrial ataxias follow maternal or, for nuclear-encoded genes such as POLG, recessive inheritance and characteristically combine ataxia with epilepsy, neuropathy, ophthalmoplegia or stroke-like episodes [21,24].

Ataxia syndrome	Genetic / pathology	Typical age at onset	Chief clinical features	Characteristic features	Geographic / prevalence notes
POLG-related ataxia (e.g. MIRAS, SANDO, SCAE)	POLG mutations (nuclear polymerase-γ), usually AR → mtDNA instability	Childhood to adult (variable)	Progressive cerebellar ataxia, polyneuropathy, epilepsy, ophthalmoplegia/ptosis; migraine or stroke-like episodes, basal ganglia features	Secondary multiple mtDNA deletions. Overlaps MIRAS, SANDO (sensory ataxic neuropathy, dysarthria, ophthalmoparesis) and SCAE (ataxia with epilepsy). Lactate often elevated	Worldwide; relatively frequent among mitochondrial ataxias in northern Europe
MELAS	mtDNA point mutations (e.g. MT-TL1), maternal	Childhood or teens	Stroke-like episodes with seizures, hemiparesis, cortical blindness; progressive encephalopathy, myopathy, lactic acidosis, migraine, hearing loss; ataxia from accumulated brain injury	Recurrent "non-vascular" strokes before age 40; ragged-red fibres on muscle biopsy; lactic acidosis crises	Global; one of the more common mitochondrial encephalopathies
MERRF	mtDNA mutations (e.g. MT-TK), maternal	Childhood or adolescence	Triad of myoclonus, generalised epilepsy and cerebellar ataxia; muscle weakness, hearing loss, later dementia and exercise intolerance	Ragged-red fibres; variable severity depending on heteroplasmy; tremor and lipomatosis common	Rare but classic mitochondrial syndrome, worldwide distribution
NARP	mtDNA MT-ATP6 mutation (e.g. 8993T>G), maternal	Childhood	Peripheral neuropathy and cerebellar ataxia with balance problems; retinitis pigmentosa with night blindness and field loss; developmental delay	Triad of neuropathy, ataxia and vision loss; intermediate mutant load gives NARP, high load Leigh syndrome; strong genotype–phenotype	Rare; reported worldwide

			and seizures may occur	correlation	
Kearns–Sayre syndrome (KSS)	Large mtDNA deletions (usually sporadic)	Before age 20	External ophthalmoplegia, pigmentary retinopathy and cerebellar ataxia; heart block, short stature, hearing loss, diabetes	Defined by onset <20, PEO, retinal degeneration and ataxia; elevated lactate; classic mitochondrial multisystem disorder	Very rare; sporadic cases globally
Leigh syndrome	mtDNA or nuclear mutations (e.g. MT-ATP6, SURF1); maternal or AR	Infancy or early childhood	Developmental regression, hypotonia, feeding difficulty, lactic acidosis; seizures, ophthalmoparesis, respiratory dysfunction, ataxia; symmetric brainstem/basal ganglia lesions on MRI	Rapidly progressive infantile mitochondrial encephalopathy; often fatal by age 2–3; milder alleles overlap with NARP phenotype	Rare; seen worldwide, often in consanguineous families or specific founder groups

Diagnostic considerations differ between adults and children in their pre-test probabilities and in the balance between urgent acquired-cause exclusion and genetic/metabolic testing, as summarised in Table 5 [19,20,35].

Table 5. Adult versus paediatric diagnostic considerations.

Feature	Adult-Onset Ataxia (≥18 years)	Paediatric/Adolescent Ataxia
Common Aetiologies	Degenerative (sporadic or genetic) ataxias common: SCAs (if family history), idiopathic degenerative ataxia, MSA-C. Acquired causes: stroke, multiple sclerosis, tumour, chronic alcohol or toxin exposure, vitamin deficiencies, normal pressure hydrocephalus (gait apraxia can mimic ataxia). Autoimmune causes (e.g. late-onset gluten ataxia, paraneoplastic) also in differential.	Genetic/metabolic disorders predominate in persistent ataxias: e.g. Friedreich's ataxia (teens), ataxia-telangiectasia (childhood), mitochondrial diseases, inborn errors of metabolism. Acute ataxias often post-infectious cerebellitis or toxin ingestion. Tumours (posterior fossa neoplasms like medulloblastoma) are an important consideration in children.
Onset and Course	Often insidious onset for degenerative/genetic cases (months–years of progression). Acute onset in stroke or metabolic insult. Adults may not seek care until ataxia causes falls or impacts work.	Can be acute (post-viral cerebellitis presents over hours/days, often in toddlers after varicella or other virus) or subacute/chronic for hereditary cases. Developmental regression can be a clue (child losing milestones = think metabolic/genetic). Parents often notice gait abnormality early.
Family History	A three-generation family history is crucial. Adult hereditary ataxias may present later in life, so a negative family history does not exclude a genetic cause (some SCAs manifest after age 40, and new mutations or non-paternity can occur). Many adult patients are sporadic.	Many childhood ataxias are recessive or X-linked; a family history of early sibling deaths, parental consanguinity, or known genetic illness is a red flag. A detailed prenatal and birth history is also relevant (to consider congenital causes, perinatal injury).
Diagnostic Workup Emphasis	Extensive workup for acquired causes in sporadic adult cases: brain MRI for stroke or atrophy, serum studies for inflammation, vitamin levels, thyroid, etc. Genetic testing (panels or exome) if no cause found and suspicion for hereditary ataxia (yield of genetic testing in adult sporadic ataxia is ~10–20% in some studies). Cancer screening if paraneoplastic ataxia suspected (age-appropriate screenings, CT	In children, metabolic and genetic testing is front-loaded when history suggests a chronic progressive ataxia (e.g. serum amino acids, lactate, lysosomal enzyme assays, genetic panels) since many causes are treatable metabolic disorders. Brain MRI is critical to identify tumours or leukodystrophies. For acute ataxia in kids, workup may be minimal if a post-infectious cerebellitis is likely (MRI normal, child improving). Lumbar puncture may be

	chest/abdomen in smokers, etc.).	considered to rule out acute encephalitis or ADEM.
Diagnostic Criteria Sets	No single unified set; diagnosis is clinical. However, specific disorders have criteria: e.g. MSA (2022 revised criteria require a combination of autonomic failure plus cerebellar or parkinsonian signs, with supportive features on imaging) – a probable MSA-C diagnosis in an adult requires cerebellar syndrome plus autonomic dysfunction (orthostatic BP drop or urinary retention). Primary Autoimmune Cerebellar Ataxia (PACA) has proposed criteria: subacute onset of idiopathic ataxia, evidence of CNS autoimmunity (e.g. MRI inflammation or CSF oligoclonal bands or relevant autoantibodies), and exclusion of other causes.	Paediatric ataxia disorders often have established clinical criteria: e.g. Ataxia Telangiectasia – progressive ataxia in a child with oculocutaneous telangiectasias and immunodeficiency (elevated alpha-fetoprotein is a diagnostic marker). Acute cerebellar ataxia of childhood is diagnosed clinically by acute onset post-viral ataxia in a toddler with normal neuroimaging. Opsoclonus–myoclonus syndrome in young children is recognised by chaotic eye movements, myoclonus and ataxia, often paraneoplastic (neuroblastoma).
Prognosis considerations	Adults with degenerative ataxias have variable prognosis: some SCAs have slower course (SCA6 patients may have near-normal lifespan with aid of devices), whereas MSA has poor prognosis (median 6–10 years survival). The ability to manage falls risk and complications (aspiration from bulbar dysfunction) factors into outcomes.	Many childhood ataxias are life-shortening (e.g. AT leads to early adult mortality due to cancer or lung infections; FRDA patients often die by middle age from cardiomyopathy). However, some childhood ataxias (post-infectious, toxin) have excellent prognosis with full recovery. Early diagnosis facilitates therapy (physiotherapy, immunoglobulin for AT, etc.).

Standardised ataxia scales (SARA, ICARS) quantify severity and progression across both age groups, and consensus genetic-testing frameworks help decide when to deploy extensive panels — typically after acquired-cause exclusion in adults, or earlier in children with suggestive metabolic features [36,37,45]. The diagnostic endpoint is either a specific disorder or a defensible syndromic category (acquired, genetic or idiopathic) that directs management.

□ **Clinical Pearl:** Two recently characterised repeat-expansion disorders have rewritten the 'idiopathic' category: CANVAS (biallelic RFC1) and the FGF14 GAA expansion (SCA27B). Both are common, both are late-onset, and both are testable — actively consider them before settling on ILOCA [17,18].

V. Investigations and the Role of Imaging

Investigation is guided by onset tempo, age, family history and associated features, and is best framed as a tiered approach (Figure 7) [4,34]. MRI of the brain (with spine where a myelopathic or FRDA pattern is suspected) is the single highest-yield test: it identifies structural lesions, demonstrates the pattern and distribution of atrophy, and can show signal change in inflammatory or paraneoplastic disease. In MSA-C the MRI may show the 'hot cross bun' pontine sign and middle-cerebellar-peduncle change, while marked cerebellar atrophy with preserved brainstem favours a pure cerebellar SCA [12,27].

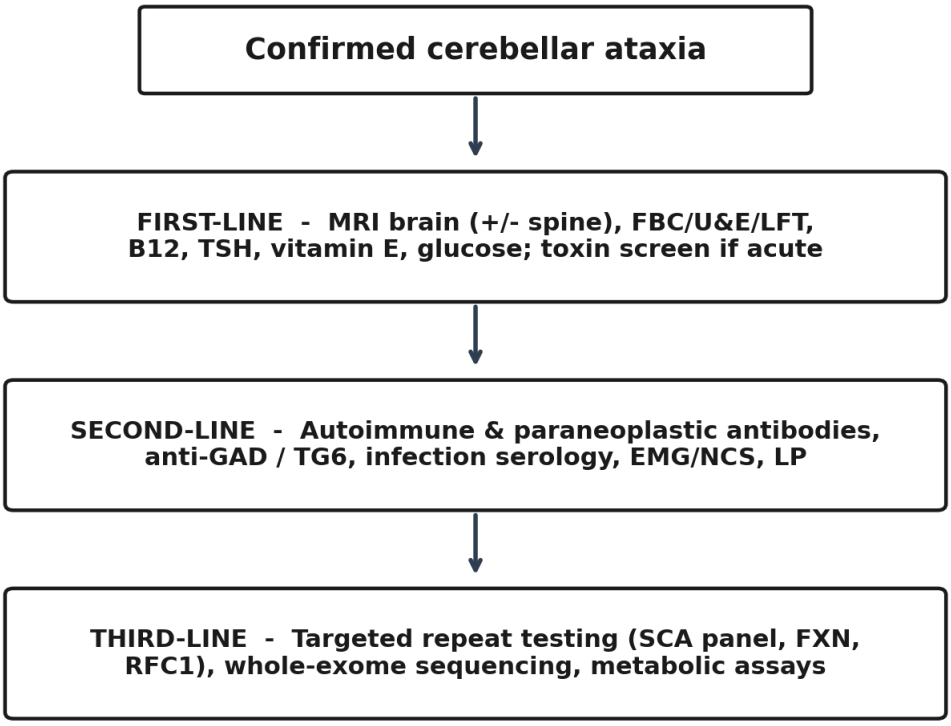

Figure 7. Tiered investigation of confirmed cerebellar ataxia.
Source: Adapted from Ashizawa and Xia [4] and de Silva et al. [34].

Table 6. Key investigations in cerebellar ataxia, with rationale and indication.

Test / Investigation	Purpose	When to Order
Neuroimaging (MRI of brain ± spine)	High-yield test to identify structural lesions or atrophy. MRI can reveal ischemic or haemorrhagic stroke in acute ataxia, tumours (posterior fossa masses), or characteristic cerebellar atrophy patterns in degenerative ataxias. Some ataxias have MRI signatures (e.g. MSA shows pontine "hot cross bun" sign; FXTAS shows middle cerebellar peduncle T2 hyperintensities). Spinal MRI if concern for cervical spondylosis or vitamin B12 deficiency myelopathy contributing to ataxia.	All patients with unexplained ataxia, unless contraindicated. MRI is recommended in essentially every ataxia workup, even if onset is chronic, to exclude lesions and assess cerebellar volume. In acute presentations, urgent MRI/CT distinguishes stroke or haemorrhage (MRI preferred for posterior fossa).
Routine Blood Tests (FBE, U&E, LFT, BSL, B12, folate, TSH)	General screen for metabolic or systemic contributors. Vitamin B12 deficiency can cause combined cerebellar and peripheral ataxia. Thyroid dysfunction (esp. hypothyroidism) can cause gait impairment and is associated with autoimmune ataxia (Hashimoto's encephalopathy). These tests also provide baseline for further investigations.	All new ataxia patients as initial screen – particularly B12 in vegans or elderly, and glucose (hypoglycaemia can cause acute ataxia). TSH for metabolic encephalopathy. Liver and renal tests before considering medications or if suspect toxin accumulation.
Inflammatory Markers (ESR, CRP) and Autoimmune Panel (ANA, ENA, dsDNA, etc.)	Indicators of systemic inflammation or connective tissue disease. Ataxia can rarely be initial presentation of lupus, Sjögren's syndrome, or systemic	When subacute ataxia or multi-system autoimmune signs are present (e.g. concurrent arthritis, sicca symptoms suggesting Sjögren). Also reasonable in

	vasculitis affecting the CNS. ANA and specific antibodies (e.g. SSA/SSB for Sjögren) are checked if an immune-mediated cerebellopathy is possible.	any idiopathic subacute ataxia to screen for Autoimmune GFAP astrocytopathy or other CNS autoimmune disorders (which often have high inflammatory markers).
Paraneoplastic Neurological Panel (serum ± CSF)	Detection of onconeural antibodies associated with paraneoplastic cerebellar degeneration (e.g. anti-Yo, Hu, Ri, CV2, Ma2, Tr, amphiphysin). These suggest a remote cancer triggering the ataxia. Some newer antibodies (e.g. anti-mGluR1, ITPR1) indicate idiopathic autoimmune ataxia.	In subacute progressive ataxia especially if risk factors for cancer (age >50, smoking, weight loss). If any red flags (e.g. co-existing neuropathy, myoclonus, or radiculopathy), send panel early. Paraneoplastic ataxias often have a rapidly progressive course over weeks. Send both serum and CSF, if possible, as some antibodies (e.g. anti-Hu) may be higher titre in CSF.
Vitamin E level (± other vitamins)	Low vitamin E causes Ataxia with Vitamin E Deficiency (AVED), a treatable mimicker of Friedreich's ataxia. Low thiamine (B1) causes Wernicke's encephalopathy (ataxia with confusion and eye palsies). Low B12 causes sensory ataxia. Copper studies if Wilson's disease considered (though Wilson's usually presents with dystonia, it can cause gait ataxia).	In young patients with Friedreich-like picture but negative frataxin test, check vitamin E. Always check B1 in alcoholic or malnourished patients (and empirically treat if Wernicke's suspected). B12 for anyone with peripheral neuropathy signs or dorsal column loss. Copper studies in adolescents or young adults with any movement disorder (Kayser-Fleischer rings on exam would prompt this).
Genetic Testing (gene panels, whole-exome or specific gene tests)	To confirm a hereditary ataxia. Panels can test dozens of ataxia-related genes (SCAs, FRDA, AOA, etc.) simultaneously. For certain classic phenotypes, targeted tests: e.g. frataxin GAA repeat test for Friedreich's, FRAGILE X (FMR1) test for FXTAS in older men with tremor/ataxia, SCA repeat panel (SCA1,2,3,6,7, etc.), and RFC1 repeat test for CANVAS. Whole exome sequencing if panel is negative and suspicion remains high.	Strongly indicated if: (a) Positive family history of ataxia; (b) Sporadic ataxia onset < 40 (many idiopathic degenerative cases are older, so early onset hints at genetic); (c) Any other clues (e.g. telangiectasia, vision loss, cognitive decline) pointing to specific inherited syndromes. In late-onset sporadic cases, genetic yield is lower but still considered after other tests if no cause found. Counselling is recommended before testing, as results can have implications for family members.
Lumbar Puncture (CSF analysis)	To evaluate for CNS infection or inflammation (e.g. viral encephalitis, autoimmune encephalitis, prion disease). In ataxia, CSF can identify oligoclonal bands (supporting MS or other autoimmune causes) or 14-3-3 protein (marker of prion disease like CJD). Also allows paraneoplastic antibody testing in CSF, and cytology for malignant cells (if carcinomatous meningitis suspected).	Acute or subacute ataxia of unclear cause, especially if encephalopathy or fever is present (rule out infectious cerebellitis). Also, if MS or CNS inflammation is in differential (look for oligoclonal bands). Do in rapidly progressive ataxia where CJD is a concern (14-3-3 and RT-QuIC prion test). In new-onset ataxia with cancer history, LP helps exclude leptomeningeal metastases.
Electrodiagnostic Studies (EMG/NCS)	Nerve conduction study (NCS) and electromyography (EMG) detect peripheral neuropathy or motor unit disease. Many ataxias have an associated neuropathy (e.g. Friedreich's, CANVAS, Vitamin E def., Sjögren, Miller Fisher variant of GBS). Sensory NCS can confirm large-fibre sensory loss consistent with sensory ataxia. EMG can identify myopathic changes in disorders like hypothyroid myopathy that might mimic ataxia gait.	When exam suggests peripheral neuropathy (areflexia, loss of vibration sense) or if diagnosis could be a neuropathy variant (e.g. Miller Fisher syndrome causes ophthalmoplegia, ataxia, areflexia – essentially a peripheral process). Also, in chronic ataxia of undetermined cause, an EMG/NCS is often done to ensure a subtle neuropathy isn't missed.
Vestibular Testing (videonystagmography, head impulse test,	To assess the inner ear and vestibular function. An abnormal head impulse test or caloric test indicates peripheral	If there is prominent vertigo or head motion sensitivity in the ataxia patient, or oscillopsia (suggesting bilateral

audiometry)	vestibular loss, which can cause or contribute to ataxia (vestibular ataxia). Audiometry is helpful since some genetic ataxias have hearing loss (e.g. AOA2), and Meniere's disease (vestibular disorder) can present with imbalance.	vestibulopathy). Also, if clinical exam suggests a vestibular component (e.g. corrective saccades on head impulse test). These tests distinguish peripheral vestibular causes from central cerebellar causes of nystagmus and gait instability.
Cardiac evaluation (ECG, Echo)	Certain ataxias involve the heart (Friedreich's ataxia frequently causes hypertrophic cardiomyopathy). MSA can cause cardiac autonomic failure (arrhythmias, blood pressure lability). Cardiac monitoring can reveal arrhythmias (e.g. AF in older patients on ataxia meds like 4-aminopyridine).	Baseline ECG in known Friedreich's or if considering it. Echo if signs of cardiomyopathy (dyspnoea, murmurs) in FRDA or other metabolic disorders. In MSA, autonomic testing (including BP and heart rate variability) is often done; a tilt-table test might be warranted for suspected neurogenic orthostatic hypotension.

First-line testing pairs MRI with basic bloods (including B12, thyroid function and, where relevant, vitamin E), adding urgent toxin and thiamine work-up in the acute setting. Second-line testing addresses immune, paraneoplastic and infective causes, with nerve-conduction studies and lumbar puncture where indicated. Third-line testing comprises targeted repeat assays (SCA panel, FXN, RFC1) and whole-exome sequencing, plus specialised metabolic assays in the young [4,34]. Timeliness is decisive for treatable disease — prompt recognition of Wernicke encephalopathy, autoimmune cerebellitis, vitamin deficiency and Wilson disease prevents irreversible injury [34].

VI. Differential Diagnosis

The differential is broad and includes both the many causes of true cerebellar ataxia and the conditions that mimic it. The first discrimination is between cerebellar, sensory and vestibular ataxia. Sensory ataxia from dorsal-column or large-fibre neuropathy produces a Romberg-positive, vision-dependent unsteadiness without dysarthria or nystagmus; vestibular ataxia produces directional veering with vertigo and a unidirectional, fixation-suppressed nystagmus; cerebellar ataxia produces incoordination independent of vision, with dysarthria, dysmetria and gaze-evoked or downbeat nystagmus [3,32,34].

Table 7. Differential diagnosis of the patient presenting with 'ataxia'.

Diagnosis or Category	Distinguishing Features
Cerebellar Ataxia (true)	Presents with incoordination independent of sensory input. Gait is wide-based and unsteady even with eyes open. Often accompanied by dysarthria, nystagmus, and limb dysmetria. Romberg test: typically, negative or only mildly worse with eyes closed (because balance is impaired even with visual feedback). There may be direction of fall towards side of lesion (in unilateral cerebellar lesions).
Sensory Ataxia (proprioceptive loss)	Caused by dysfunction of dorsal columns or peripheral nerves (e.g. neuropathy, dorsal column disease like B12 deficiency or tabes dorsalis). Key features: marked worsening of balance when visual input is removed – Romberg test strongly positive (fair balance with eyes open, but unable to stand with eyes closed). Gait may involve stomping steps (to increase proprioceptive feedback) and patients often watch their feet while walking. Limb coordination may improve with visual guidance. Absent ankle reflexes and loss of vibration sense are common (if peripheral neuropathy). No true dysarthria or nystagmus (cerebellum itself is intact).
Vestibular Ataxia (peripheral or central vestibular dysfunction)	Primarily a balance disorder due to vestibular system lesions. Vertigo is prominent, often with nausea and nystagmus (especially acute vestibular disorders). Gait ataxia tends to veer to one side. In peripheral vestibular lesions (e.g. vestibular neuritis), patients may fall towards the affected ear, have unidirectional nystagmus that suppresses with fixation, and often improve with time (central compensation). Head impulse test is positive in peripheral causes. Romberg: may be positive, often with fall to one side, but not purely vision-dependent (eyes open may also be unsteady). In central vestibular lesions (e.g. lateral medullary stroke affecting vestibular nuclei or cerebellar peduncle), there is often concurrent cerebellar ataxia and other brainstem signs (diplopia, sensory loss in a crossed pattern, etc.).
Parkinsonian Gait	Gait impairment in Parkinson's disease or atypical parkinsonism (PSP, etc.)

	can be confused with ataxia. Parkinsonian gait is characterised by narrow-based, shuffling steps, stooped posture, and difficulty initiating gait (start hesitation, freezing). Turns are en bloc. There is no staggering or erratic foot placement as in ataxia; instead, steps are small and patient may festinate (speed up involuntarily). Arm swing is reduced (whereas in pure ataxia arm swing is preserved or exaggerated). Rigidity and bradykinesia on exam distinguish this from cerebellar ataxia. Note: MSA can have both parkinsonism and cerebellar signs; in those cases, both features may coexist (this is an MSA clue).
Normal Pressure Hydrocephalus (NPH)	Causes a classic triad of gait disturbance, cognitive decline, and urinary incontinence. The gait disorder is sometimes called “gait apraxia” or “frontal ataxia”: patients have a broad stance and difficulty lifting their feet (“magnetic” gait as if feet stuck to floor) with short steps. Unlike true cerebellar ataxia, truncal balance can be quite poor, but limb coordination (finger-nose) may be relatively preserved. Patients often have slowed cognitive processing. Brain imaging (enlarged ventricles) helps distinguish NPH. NPH gait tends not to have the weaving, drunken quality of cerebellar gait, but the broad base can cause confusion.
Stroke in motor cortex or corticospinal tract	Leg weakness or spasticity can cause unsteadiness that mimics ataxia, but primary issue is motor power or tone. Corticospinal (spastic) gait: stiff-legged with scuffing of feet, often unilaterally (if hemiparesis) or scissoring (if paraparesis). Hyperreflexia and Babinski signs will be present. Coordination tests may appear slow due to weakness, but not irregular as in ataxia. Important because a mild hemiparesis can be mistaken for ataxia if not carefully examined (look for pronator drift, increased tone).
Functional (Psychogenic) Gait Disorder	Some patients have gait imbalance without a neurological disease, often due to functional neurological disorder. Clues include inconsistency (dramatic sway but ability to catch self, or normal tandem walking when distracted), exaggerated effort and uneconomical postures, improvement with encouragement or when unaware of observation. In functional gait, falls are rare (despite apparent near falls) and there may be excess arm waving. Hoover’s test or other signs of functional weakness might coexist. No nystagmus or limb ataxia on exam (or if present, inconsistent). Diagnosis of exclusion, but important to recognise to avoid unnecessary invasive tests.

Further mimics include cervical spondylotic myelopathy (proprioceptive ataxia with spasticity and Lhermitte phenomenon), drug-induced ataxia (sedating anti-epileptics are the classic culprits, reversible on withdrawal), and, in children, ingestion and basilar migraine [27,34]. The combination of history and examination usually narrows the field decisively: a positive Romberg with distal sensory loss directs work-up toward neuropathy or B12 deficiency, whereas dysarthria and limb dysmetria confirm cerebellar localisation.

□ **Clinical Insight:** In the acute vestibular syndrome the discriminating question is not 'how severe is the vertigo' but 'is the head-impulse test normal'. A normal HIT in a patient with spontaneous nystagmus is a central sign and shifts the differential toward cerebellar infarction [27,32].

VII. Management — Rehabilitation and Pharmacotherapy

Management is two-pronged: treat the underlying cause where possible, and mitigate symptoms and disability where it is not. Many ataxias are incurable, but structured supportive care meaningfully improves function and safety, and should begin at the time of diagnosis rather than being deferred [3,39]. Figure 8 summarises the overall strategy.

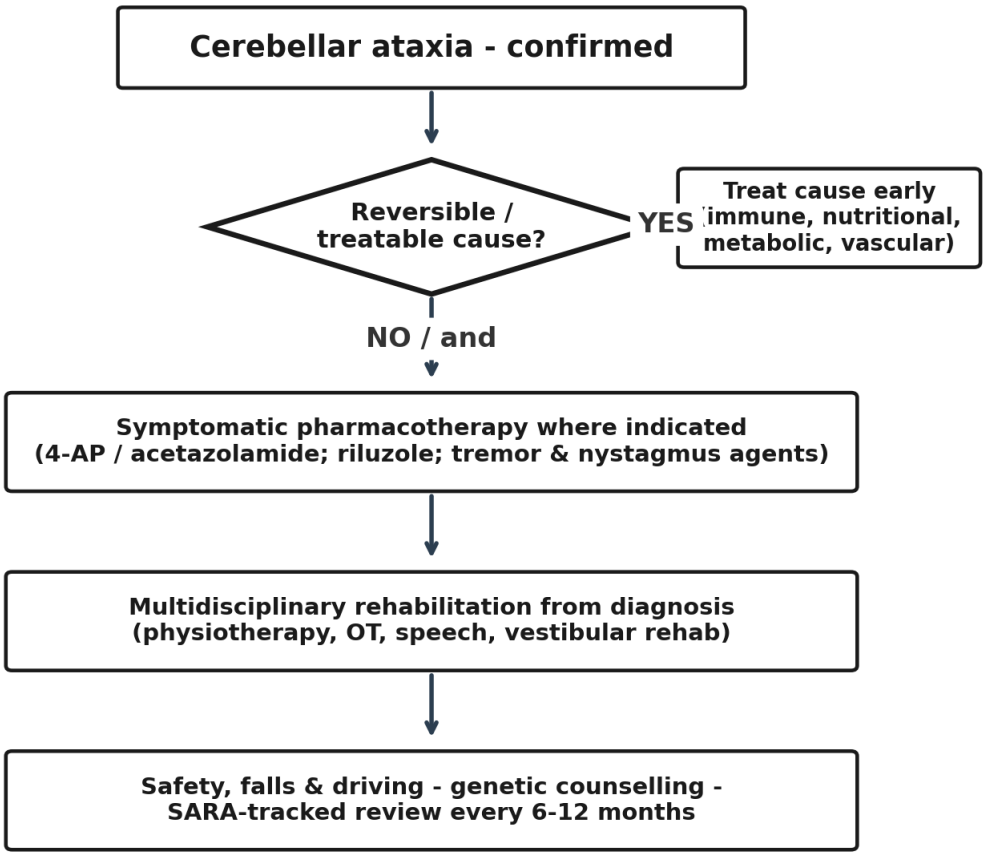

Figure 8. Management algorithm for confirmed cerebellar ataxia.
Source: Adapted from Zesiewicz et al. [39] and Mitoma et al. [30].

Non-pharmacological management

Multidisciplinary rehabilitation is the cornerstone. Intensive coordinative physiotherapy targeting balance, gait and trunk control produces measurable, durable improvement in degenerative cerebellar disease: controlled trials of four-week intensive programmes show clinically relevant gains in SARA and functional independence that persist for months [43,44]. Occupational therapy addresses fine-motor function and home safety, speech therapy addresses dysarthria and dysphagia, and vestibular rehabilitation adds benefit where there is a vestibular component such as CANVAS or coexisting vestibulopathy [18,39]. Assistive devices (broad-based or wheeled aids, ankle-foot orthoses, weighted utensils), falls-prevention and home modification, nutritional and psychosocial support, and strict avoidance of alcohol complete the non-pharmacological package [39].

Symptomatic pharmacotherapy

No drug reverses cerebellar neurodegeneration, but several reduce specific symptoms. The 2018 American Academy of Neurology systematic review remains the principal evidence synthesis [39]. Aminopyridines are the most useful class for vestibular practice: 4-aminopyridine reduces attack frequency in episodic ataxia type 2 in randomised data and improves downbeat and upbeat nystagmus and gaze stability in degenerative cerebellar disease [40,41]. Acetazolamide is long established for EA2 and reduces attack frequency [40]. Riluzole improved ataxia ratings in randomised trials in mixed and hereditary ataxias and may be trialled in progressive disease with monitoring of liver enzymes [42]. Other agents — gabapentin or memantine for downbeat nystagmus, clonazepam or baclofen for cerebellar tremor and myoclonus, and selected second-line options — are used selectively and individually titrated [39,41].

Table 8. Symptomatic pharmacological agents in cerebellar ataxia.

Drug (Class)	Mechanism / Target	Dose Range	Monitoring & Cautions
Acetazolamide (carbonic anhydrase inhibitor)	Modulates neuronal pH and ion balance; in EA2 thought to improve cerebellar ionic homeostasis to prevent attacks.	250–1000 mg per day (e.g. 250 mg BID up to QID). Titrate to response.	Monitor serum bicarbonate (can cause metabolic acidosis), electrolytes, and renal function. Watch for paresthesias, kidney stones. Clinical response in days; if none in 1–2 months for EA2, reconsider.
4-Aminopyridine (4-AP) (K^+ channel blocker)	Prolongs action potentials and enhances residual Purkinje cell firing, improving cerebellar output (also improves ocular stability in nystagmus).	Immediate-release: Start 5 mg TID, titrate up to ~10 mg QID (max ~40 mg/day) as tolerated. Sustained-release form (dalfampridine) 10 mg BID often used.	Contraindicated in history of seizures (lowers threshold). Monitor for dizziness, nervousness – toxicity can cause seizures or arrhythmias. If no benefit at max tolerated dose in 1 month, likely ineffective.
Riluzole (glutamate release inhibitor)	Reduces glutamatergic transmission; in ataxia, may protect against glutamate excitotoxicity in cerebellum.	50 mg twice daily. (Used off-label for ataxia; approved dose from ALS.)	Monitor LFTs monthly for 3 months, then quarterly (risk of hepatic injury). Side effects: fatigue, nausea. Not to be used in severe liver or renal disease. Re-assess at 12 months for efficacy.
Gabapentin (antiepileptic)	Unclear in ataxia; may reduce cerebellar tremor and nystagmus by modulating GABA release.	300 mg up to 1200 mg daily (divided TID). Often 900 mg/day used for nystagmus.	Sedation and dizziness common – titrate slowly. Adjust dose in renal impairment. Can be combined with memantine for nystagmus if needed.
Clonazepam (benzodiazepine)	Enhances $GABA_a$ activity; suppresses cerebellar tremors, myoclonus.	0.25 mg HS to 1–2 mg TID (tolerance may develop). Use lowest effective dose.	Causes sedation, cognitive slowing, risk of dependence. Caution in elderly (falls). Avoid alcohol use. Taper slowly if discontinuing to prevent withdrawal seizures.
Baclofen (muscle relaxant)	GABA_B agonist; reduces muscle tone and perhaps dampens oscillations in tremor.	5 mg TID start, titrate to 20 mg TID if needed. Can also use intrathecal pump for severe spasticity.	Side effects: drowsiness, weakness (can paradoxically worsen gait if over-relaxes antigravity muscles). Do not stop abruptly (risk of withdrawal hallucinations).
IV Methylprednisolone (corticosteroid)	Broad immunosuppressive and anti-inflammatory effects to treat autoimmune cerebellar inflammation.	Typical induction: 1 g IV daily for 3–5 days, then switch to oral prednisone taper (e.g. 60 mg daily, taper over 2–3 months). For milder cases, high-dose oral (e.g. prednisone 1 mg/kg) could be used.	Monitor blood glucose (can cause hyperglycaemia), blood pressure, mood changes. Long-term use needs bone protection (calcium, vitamin D) and PPI for gastric protection. Watch for rebound ataxia as

			steroids taper – may indicate need for longer course or second-line immunotherapy.
IV Immunoglobulin (IVIG)	Immunomodulation via pooled antibodies; likely neutralises pathogenic antibodies in immune ataxias.	2 g/kg total per cycle, given over 2–5 days (e.g. 0.4 g/kg/day ×5 days). Maintenance: 0.4 g/kg monthly if responded.	Monitor for infusion reactions (headache, hypertension), rare thrombosis or aseptic meningitis. Ensure adequate hydration and slow infusion in IgA-deficient patients (anaphylaxis risk). Expensive; justify in proven or strongly suspected immune ataxia (e.g. anti-GAD ataxia).
Gluten-free Diet (for gluten ataxia)	Eliminates dietary gluten to reduce immune trigger. Can lead to stabilisation or modest improvement in gait over time in gluten ataxia (and prevents enteropathy).	Life-long strict avoidance of gluten (wheat, barley, rye). May need dietician support.	Monitor anti-gliadin or anti-TG2 antibody titres for compliance (though TG6 is more specific for neurological gluten ataxia). Neurologic improvement may take 6–12 months. Ensure nutritional adequacy of diet.
Vitamin E (tocol)	Replenishes deficient antioxidant (in AVED) to halt neuronal damage.	400–1500 IU daily, adjust to keep serum vitamin E in high-normal range. High doses split BID to improve absorption.	Monitor vitamin E levels, cholesterol (since AVED patients may have fat malabsorption). If no neurological improvement in 1 year, further progression usually at least slows. Overdose can impair clotting – watch INR if on anticoagulants.
Thiamine (Vitamin B1)	Cofactor for metabolism; treats Wernicke's and prevents Korsakoff syndrome.	Acute Wernicke's: 500 mg IV TID for 2 days, then 250 mg IV daily ×5 days, then oral 100 mg daily indefinitely.	Give before glucose in suspected Wernicke's to avoid precipitating encephalopathy. Monitor clinical response (improvement in ocular signs within days, ataxia over weeks). Essentially no overdose toxicity, but inadequate treatment leads to permanent deficits.
Chelators (Penicillamine) – for Wilson's disease	Binds excess copper, promotes excretion; reverses copper deposition in CNS that could cause ataxia.	250 mg oral TID, gradually increase to 1–1.5 g/day in divided doses. (Or use Trientine 750 mg TID as alternative.)	Monitor full blood count (risk of cytopenias), liver function, and urinary copper excretion. Ensure pyridoxine supplementation with penicillamine. Monitor neurologic status – paradoxical worsening can occur early in Wilson treatment, requiring close supervision.

The choice of agent is individualised and combinations are common (for example 4-AP for nystagmus with clonazepam for tremor). Benefit must be reviewed objectively; if there is no subjective or measurable improvement within a few months the drug should be tapered to avoid polypharmacy [39].

VIII. Disease-Modifying and Cause-Specific Treatment

A minority of ataxias are genuinely treatable, and identifying them is the highest-value act in the whole work-up. Immune-mediated ataxias warrant immunotherapy: high-dose corticosteroids are usually first-line, with IV immunoglobulin or plasma exchange in selected cases, and steroid-sparing agents such as mycophenolate or rituximab for antibody-defined disease such as anti-GAD ataxia [28,29,30]. The guiding principle is preservation of cerebellar reserve — early treatment yields better outcomes, which justifies a trial of immunotherapy in sporadic ataxia of short duration with inflammatory MRI or serological clues, after malignancy screening [30,45]. In gluten ataxia a strict gluten-free diet can stabilise or modestly improve the ataxia and is a benign intervention [16].

Nutritional ataxias are eminently treatable: vitamin-E deficiency (including AVED) with high-dose vitamin E, Wernicke encephalopathy with emergent parenteral thiamine, and B12 or copper deficiency with replacement [34,47]. Several rare metabolic ataxias have specific therapies — miglustat for Niemann–Pick C, dietary restriction and apheresis for Refsum disease, chelation for Wilson disease, and high-dose coenzyme Q10 for CoQ10-deficiency ataxia, a treatable cause worth actively excluding in young-onset disease [20,47]. For the polyglutamine SCAs and FRDA no disease-modifying drug is yet approved, although antisense-oligonucleotide and gene-targeted programmes are in trials [2,48].

MSA-C has no disease-modifying therapy; management is supportive and multidisciplinary, with attention to autonomic failure, parkinsonism and bulbar dysfunction [12,38]. Acute causes follow their own protocols — stroke pathways for cerebellar infarction, with neurosurgical decompression for space-occupying haemorrhage or oedema; steroids for severe post-infectious cerebellitis; and oncological or neurosurgical treatment for tumour [27].

□ **Important:** The treatable ataxias — immune, nutritional (thiamine, B12, vitamin E, copper), Wilson disease and CoQ10 deficiency — must be screened for in every new ataxia, because delay converts a reversible disorder into a permanent one [30,34,47].

IX. Prognosis, Recurrence and Special Populations

Prognosis depends almost entirely on aetiology. Hereditary degenerative ataxias are slowly progressive: FRDA typically begins in the teens with wheelchair dependence on average 10–15 years later, and hypertrophic cardiomyopathy is the leading cause of premature death, although modern cardiac care has extended survival [46]. Among the dominant ataxias the course varies — SCA1 and SCA2 progress faster, SCA3 over roughly two decades, and SCA6 is mild with many patients ambulant for decades and near-normal lifespan [5,6]. These are not relapsing conditions; day-to-day fluctuation with fatigue or intercurrent illness is common but does not represent true relapse [36].

MSA-C carries a poor prognosis, progressing faster than most SCAs with mean survival of roughly 6–10 years and death usually from bulbar or autonomic complications [12,38]. Immune ataxias have the most favourable outlook when treated early — gluten ataxia often stabilises on a gluten-free diet and anti-GAD ataxia may improve with immunotherapy, though residual deficit is common — whereas paraneoplastic cerebellar degeneration is typically a monophasic but devastating insult with limited recovery [28,29,30]. Post-infectious cerebellitis in children has an excellent prognosis with near-universal full recovery [19]. Episodic ataxias are, by definition, relapsing with normal interictal function in EA1 and frequently normalised attacks on acetazolamide in EA2; prophylaxis can reduce attack frequency dramatically [33,40].

Two themes recur for the vestibular physician. First, the vestibulocerebellum is integral to vestibulo-ocular reflex calibration, organised into parallel flocculus and nodulus streams for gaze stabilisation and spatial orientation (Figure 9); cerebellar disease therefore produces disequilibrium, oscillopsia and impaired gaze

stability, and acute cerebellar stroke can present as isolated vertigo indistinguishable from neuritis without a careful oculomotor examination [27,32]. Second, CCAS means cognitive and affective screening belongs in ataxia follow-up, not only motor rating [23,31].

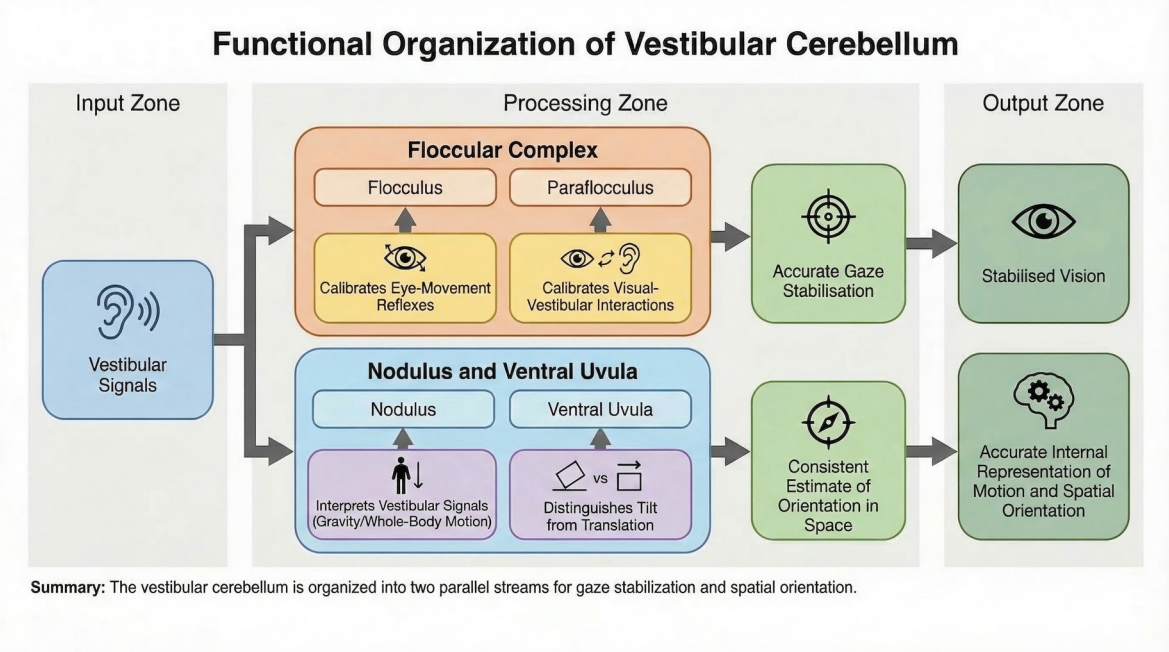

Figure 9. Functional organisation of the vestibulocerebellum — parallel input, processing and output streams for gaze stabilisation and spatial orientation.
Source: Australian Dizziness Clinics.

Genetic counselling is integral to hereditary disease — dominant ataxias carry a 50% transmission risk, predictive testing of adults requires structured pre-test counselling, and testing of asymptomatic minors for adult-onset disease is generally discouraged [35,45]. Natural-history data from consortia such as EUROSCA and the Friedreich registries now quantify yearly progression by genotype, which supports both prognostic counselling and trial design [36,48]. Prognosis should be communicated honestly but with attention to wide individual variation and to the emerging therapeutic pipeline.

X. Guidelines, Controversies and Future Directions

Several debates define the current field. The classification of idiopathic ataxia is shifting rapidly: the discovery of the RFC1 expansion in CANVAS, and subsequently the FGF14 expansion (SCA27B), has shown that many 'sporadic' late-onset ataxias have an as-yet-undiscovered genetic basis, blurring the boundary between genetic and degenerative disease and challenging the durability of the ILOCA label [13,14,17,18].

Primary autoimmune cerebellar ataxia (PACA) is a second flashpoint. International criteria were proposed in 2020, but critics note that low-titre antibodies of uncertain significance and atypical genetic or paraneoplastic disease can masquerade as PACA, so the threshold for empirical immunotherapy in sporadic ataxia remains contested [28,45]. Gluten ataxia provokes a related argument over how widely to screen and which antibody titre is diagnostic, given imperfect serological specificity [15,16]. Pharmacological practice is similarly unsettled: aminopyridines are well supported in episodic ataxia and nystagmus, but their role in progressive ataxia is empirical and off-label, and uptake of riluzole varies despite randomised data, reflecting modest effect sizes [39,40,41,42].

Looking forward, the priorities are biomarkers and disease-modifying therapy. Blood neurofilament light, quantitative oculomotor measures, volumetric and advanced MRI, and wearable gait sensors are being validated as responsive endpoints, an effort coordinated internationally by the Ataxia Global Initiative to enable adequately powered trials in these rare diseases [48]. Gene-silencing and gene-replacement strategies for the polyglutamine SCAs and FRDA are in early human testing, and the field is cautiously optimistic that the cerebellum's relative surgical accessibility will make it an early proving ground for targeted neurotherapeutics [2,48]. The vestibular physician's role across all of this is constant: recognise

the cerebellar pattern, exclude the dangerous and the treatable promptly, quantify and rehabilitate, and connect patients to genetic services and trials.

□ **Key Point:** The 'idiopathic' ataxia bucket is shrinking. Re-interrogate older sporadic diagnoses as new repeat-expansion tests (RFC1, FGF14) and immune criteria become available — a proportion will be reclassified into actionable categories [17,18,45].

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