

# Expanded *ATXN10* Alleles in Neurodegenerative Disorders: A Case Series and Review of the Literature

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## Case Report

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## Abstract

**Background:** Spinocerebellar ataxia type 10 (SCA10) is typically attributed to large intronic ATTCT repeat expansions in *ATXN10*, yet interpretation is complicated by repeat interruptions, reduced penetrance, and assay limitations.

**Cases:** We describe three patients referred for SCA10 evaluation in whom *ATXN10* repeat-primed PCR (RP-PCR) reported “>32 repeats, probable pathogenic.” Targeted long-read sequencing (LRS) identified two individuals with intermediate, mixed repeat tracts (~100–150 repeats) and one individual with a very large, highly pure ATTCT expansion (~1,127 repeats). Each phenotype was explained by an alternative disorder: spastic ataxia syndrome caused by biallelic pathogenic *FA2H* variants, full-penetrance *ATXN3* CAG expansion consistent with MJD/SCA3, and clinically established MSA-C.

**Literature Review:** We identified three studies reporting four SCA10 cases coexisting with Huntington’s disease (one case) and SCA2 (three cases).

**Conclusion:** Our three cases suggest that intermediate *ATXN10* alleles in the 100–150 range may not be pathogenic and that even very large, pure expansions may be non-pathogenic. Comprehensive genetic testing beyond RP-PCR is necessary for accurate diagnosis.

## Introduction

*ATXN10* intronic repeat expansions are associated with spinocerebellar ataxia type 10 (SCA10), an autosomal dominant disorder typically characterized with slowly progressive cerebellar ataxia, dysarthria, ocular abnormalities, nystagmus, cognitive impairment, and epileptic seizures.<sup>1</sup> Expanded ATTCT have been traditionally related to pathogenic presentations above a threshold of 850 ATTCT repeats.<sup>2,3</sup> The phenotypic variability is reported across different populations, with differences in age at onset, disease duration, and epileptic seizures frequency.<sup>1</sup> The overlap between reduced and full penetrance ranges has been described, and expanded alleles have occasionally been identified in healthy individuals and individuals with atypical or discordant neurological phenotypes.<sup>4,5</sup>

Here, we report three subjects, two harboring intermediate mixed ATTCT alleles and one with a pure expanded ATTCT allele in the *ATXN10* gene whose clinical presentations were attributable to another confirmed disorder. We compared these cases with other reports in literature, describing clinical features, disease evolution, and diagnostic methods applied for genetic confirmation using traditional repeat expansion analysis, targeted long-read sequencing (LRS) and whole-genome sequencing (WGS).

## Case Series

### Case 1

A 34-year-old woman presented with symptom onset at age 24, during late first pregnancy, initially characterized by foot dragging while walking. At 25, she developed recurrent falls, progressive dysarthria, and lower-limb cramps. There was no relevant past medical or family history (**Supplementary Figure S1**).

Neurological examination (age 25) revealed preserved orientation and cognition, with mild distal lower-limb weakness, generalized hyperreflexia, bilateral Babinski and Hoffman signs, and lower-limb spasticity. Sensory testing disclosed reduced vibration sense in the lower-limbs. Cerebellar signs included dysidiadochokinesia, limb dysmetria, and multidirectional nystagmus. Spastic gait was present with moderate impairment. The Scale for the Assessment and rating of Ataxia (SARA) was 8.5/40 and the Neurological Examination Score for Spinocerebellar Ataxia (NESSCA) 14/40, with Ashworth grade 3 spasticity reflecting increased tone and resistance to passive movement. Laboratory workup was unremarkable, including normal vitamin B12 and folate levels, negative HTLV-1 serology, normal rheumatoid factor, and normal copper studies (24-hour urinary copper and ceruloplasmin). Electromyography (EMG) showed no evidence of peripheral neuropathy. A report of brain MRI described mild bilateral cerebellar atrophy (MRI images were not available).

She was followed regularly every 3–6 months over 8 years, showing gradual progression of motor impairment with worsening spastic gait, although she remained ambulatory (SARA 15/40 at age 32) (**Video 1**). Cognitive disturbances became evident with Montreal Cognitive Assessment (MoCA 14/30), accompanied by severe depressive episodes. Symptomatic management included baclofen titrated up to 60 mg/day, anticholinergic therapy, escitalopram 30 mg/day, and ongoing physical therapy. Ataxia genetic testing panels were CAG repeats negative for SCA1 (29/30), SCA2 (22/22), MJD/SCA3 (14/22), and SCA6 (11/12). SCA10 genotyping showed an expanded allele >32 repeats by RP-PCR which was further evaluated by LRS and revealed an *ATXN10* intermediate allele mixed repeat motifs (ATTCT)<sub>92</sub>(ATTCTAT)<sub>8</sub> (**Figure 2A**). Given the lack of phenotypic concordance with SCA10, further WGS analysis identified a biallelic *FA2H* pathogenic variant, c.461G>A (p.Arg154His).

### Case 2

A 56-year-old man with no family history of ataxia (**Supplementary Figure S2**) presented with progressive neurological symptoms beginning at age 51, initially with mild dysarthria followed by gait instability and recurrent falls. By age 55, he developed prominent autonomic dysfunction, including nocturnal urinary urgency, constipation, occasional syncope episodes, and sensory disturbances. Past medical history was notable for benign prostatic hyperplasia and prior alcohol overuse. Neurological examination (age 54) revealed preserved cognition, lower-limb areflexia, moderate gait and limb ataxia, dysarthria, saccadic intrusions, and horizontal nystagmus. SARA was 17/40 and NESSCA 7/40. Laboratory workup was unremarkable including glucose, vitamin B12 and thyroid levels. Tilt-table testing demonstrated neurogenic orthostatic hypotension (BP drop from 106/71 mmHg supine to 97/56 mmHg during head-up tilt). Bladder ultrasonography showed post-void urinary residual >100 mL. Brain MRI showed cerebellar atrophy and “hot cross bun” sign (**Figure 1**), and EMG revealed mild axonal sensory polyneuropathy.

Over three years of follow-up, he exhibited progressive cerebellar ataxia (SARA 25/40) with worsening dysautonomia and emergence of mild upper-limb rigidity and bradykinesia. Diagnosed mixed anxiety-depressive disorder responded to serotonergic therapy. Last neurological examination at age 56, highlights moderate ataxic gait and limb ataxia, and bradykinesia (**Video 2**). Genetic testing was CAG negative for SCA2 (23/23) and MJD/SCA3 (24/30). RP-PCR was positive for SCA10 (>32 repeats), and LRS revealed *RFC1* (9/10 AAAAG) and *FGF14* (9/15 GAA) on normal ranges and confirmed a highly pure ATTCT expansion of 1,127 repeats (**Figure 2B**).

## Case 3

A 44-year-old mechanical engineer with a paternal history of similar symptoms (**Supplementary Figure S3**) first presented at age 30 with early-morning paroxysmal episodes characterized by brief (<1 minute) loss of consciousness, generalized tonic posturing, tongue biting, and postictal confusion lasting approximately 30 minutes, without sphincter incontinence. Six additional events occurred over the following year. Antiepileptic medications were intermittently prescribed and discontinued; recurrence at age 43 led to maintenance therapy with lamotrigine 100 mg/day. From age 37, he developed slowly progressive gait instability with recurrent falls, predominantly related to lower-limb spasticity with hyperextension posture, accompanied by mild dysarthria, dysphagia, and exercise-induced lower-limb spasms. He has required a walker since age 41. The neurological examination (age 36) showed preserved cognition, lower-limb hyperreflexia, mild cerebellar dysarthria, spastic gait with hyperlordosis and knee hyperextension, slow saccades, and horizontal nystagmus. SARA and NESSCA were 6/40 and 10/40, respectively. Laboratory studies were unremarkable except for elevated vitamin B12; thyroid function, VDRL, and folate were normal. EMG showed no neuropathy, and repeated EEGs were normal. Brain MRI demonstrated mild global cerebellar atrophy (**Figure 1**).

Over 5 years of follow-up, he showed progressive spastic ataxia (SARA 13/40; NESSCA 16/40), with partial improvement of spasticity following botulinum toxin injections and anticholinergic therapy. At the last evaluation, he required assistance to ambulate and had developed supranuclear ophthalmoplegia with strabismus, partially corrected with prism lenses. Cerebellar cognitive affective syndrome screening (CCAS) scored 91/120, including three failed tests (category switching, verbal recall, affect). Genetic testing was positive for CAG repeat expansion on *ATXN3* (23/75), as well as mixed *ATXN10* intermediate allele of 139 ATTCT repeats ((ATTCT)<sub>100</sub>(ATTCT)<sub>1</sub>(CTTCT)<sub>38</sub>) (**Figure 2C**).

## Literature Review

We conducted a literature review to identify cases of SCA10, documenting a second neurodegenerative disorder with clinical or genetic confirmation. In addition, we reviewed studies presenting intermediate *ATXN10* expansion independently of clinical phenotype.

We found three case reports of *ATXN10* expansions with a coexisting neurodegenerative disorder. In total, we found four cases with dual diagnoses of SCA10, one case together with Huntington's disease (HD),<sup>6</sup> and three cases (two families) with SCA2.<sup>7,8</sup> In all cases, *ATXN10* repeat expansion was declared above 850 repeats, as well as CAG repeat expansion in *HTT* and *ATXN2*. Curiously, all reported cases were of Latin American origin (**Table 1**).

Previous reports suggest that *ATXN10* alleles in the roughly 280–850 repeat range may show reduced penetrance, although most early studies relied on repeat length estimates using Southern blot rather than LRS of the repeat composition. In one report, a patient with early-onset ataxia carried an unusual 280-repeat allele, while the asymptomatic mother harbored the same 280-repeat expansion containing multiple ATGCT interruptions proximally and an ATTCTAT heptanucleotide interruptions distally, with a long internal stretch of pure ATTCT.<sup>2</sup> Another study described affected individuals with 400 and 750–760 repeats, whereas unaffected relatives carried 360–370 repeats, supporting a reduced-penetrance zone.<sup>9</sup>

In a Brazilian family, a symptomatic woman had an expansion of about 850 repeats, however, six asymptomatic relatives carried similar-sized alleles.<sup>10</sup> More recently, our group showed with amplification-free LRS that repeat structure may be as important as repeat size: pure ATTCT expansions may be less pathogenic or even nonpathogenic, whereas interrupted expansions appear more strongly associated with SCA10 phenotypes.<sup>4,5,11</sup> In addition, we also identified 3.7% of intermediate *ATXN10* alleles (> 32 repeats) in Mestizo population, and 9.9% in Amerindian population in a Peruvian healthy cohort, complicating accurate diagnosis of SCA10 in some populations.<sup>12</sup>

## Discussion

Our case series highlights two recurring interpretive challenges in *ATXN10* repeat testing: (i) intermediate expansions in the ~100–150 repeat range and (ii) large, pure ATTCT expansions. In two subjects, LRS demonstrated intermediate alleles with mixed motif composition, falling in the range often encountered during clinical evaluation of ataxia but not sufficient to explain disease. Another case with highly pure ATTCT expansion over 1000 repeats fulfilling Multiple System Atrophy-cerebellar type (MSA-C) diagnosis, suggesting either a coexisting disorder or an atypical SCA10 case. Critically, all individuals had clinical phenotypes and neurological findings inconsistent with classic SCA10, and their presentations were better explained by alternative etiologies after additional testing (biallelic *FA2H* in case 1; clinically established MSA-C in case 2, and full-penetrance *ATXN3* expansion in case 3). These findings suggest that *ATXN10* alleles around 100–139 repeat range are unlikely to be pathogenic on their own and should not be considered as a stand-alone molecular diagnosis, while coexistent neurodegenerative diseases or atypical SCA10 cases might be associated with highly pure ATTCT expansions.

The case of sporadic early adult-onset spastic ataxia syndrome (case 1) initially appeared genetically compatible with SCA10 due to the detection of an ATTCT expansion in *ATXN10* by RP-PCR (>32 repeats). However, the clinical phenotype was not concordant with classical SCA10 where cerebellar ataxia, neuropathy, variable frequency of seizures are typical features.<sup>1</sup> Moreover, LRS clarified the repeat structure as a complex (ATTCT)<sub>92</sub>(ATTCTAT)<sub>8</sub> configuration, consistent with an intermediate expansion range not clearly established as pathogenic for SCA10.<sup>9</sup> WGS subsequently identified a homozygous missense variant (p.R154H) within the *FA2H* gene. This biallelic pathogenic variant typically cause Spastic Paraplegia 35 (SPG35), also known

as Fatty Acid Hydroxylase-associated Neurodegeneration (FAHN), characterized by early onset, spastic paraparesis, ataxia, dystonia, and cognitive decline.<sup>13</sup> Although most reported cases present in childhood with rapidly progressive disease and leukodystrophic changes, atypical adult-onset forms have been described, frequently with slower progression, thinning of the corpus callosum, and occasionally without leukodystrophy.<sup>14,15</sup> Our case presents with early adult onset, progressive spastic ataxia syndrome, and cognitive decline, which is consistent with this phenotypic spectrum. Approximately 70% of reported SPG35 cases coexist with cerebellar ataxia, reinforcing the inclusion of SPG35 on spastic ataxia syndromes.<sup>16</sup> The R154H missense variant has been previously reported, as well as the location on potential mutational hotspots.<sup>15</sup> Functional studies support a loss-of-function disease mechanism.<sup>14,17,18</sup> Furthermore, genotype–phenotype correlation studies suggest that variants retaining partial residual activity may be associated with milder or later-onset presentations of SPG35/FAHN.<sup>18,14,19</sup> The absence of family history, together with a homozygous pathogenic *FA2H* variant, and a phenotype highly consistent with SPG35 supports a diagnosis of late-onset SPG35. The intermediate *ATXN10* expansion is best interpreted as a coincidental finding without diagnostic significance.

The MSA-C case (case 2) presented with late-onset cerebellar ataxia and marked autonomic dysfunction. Genetic testing unexpectedly revealed a very large *ATXN10* expansion (~1,127 repeats), composed almost entirely of uninterrupted ATTCT motifs. Despite the expansion size, the phenotype was not suggestive of SCA10, and there was no family history of ataxia, epilepsy, or related neurological disease. Cerebellar syndrome, alongside orthostatic hypotension, post-void urinary residual volume >100 mL, and “hot cross bun” sign on MRI fulfilled the 2022 Movement Disorder Society criteria for clinically established MSA-C.<sup>20</sup> Given the very late onset and the coexistence of neuropathic features, alternative diagnoses such as secondary neuropathies, CANVAS and SCA27B were also considered and systematically excluded through appropriate clinical and molecular approaches. Thus, two non–mutually exclusive interpretations should be considered: (i) an atypical SCA10 presentation mimicking MSA-C, although lacking the cardinal clinical hallmarks of SCA10; or (ii) a typical MSA-C with a coincidental large but clinically silent *ATXN10* pure expansion. This case underscores that repeat size alone might be insufficient for diagnostic attribution: interruptions and complex motif architectures have been implicated in phenotypic variability, and the presence of a large, uninterrupted (pure) ATTCT tract may not confer pathogenicity in some individuals. Thus, an “expanded” *ATXN10* allele, particularly if pure, should be interpreted cautiously, in the context of phenotype, segregation, and orthogonal testing.

The adult-onset MJD/SCA3 case (case 3) was found to harbor a concomitant intermediate *ATXN10* expansion allele. The clinical presentation characterized by progressive ataxia with mild lower-limb spasticity, is consistent within the recognized spastic ataxia endophenotype of MJD/SCA3, in which pyramidal signs may coexist with cerebellar dysfunction.<sup>21</sup> Although seizures are uncommon in MJD/SCA3, they may or may not be part of the broader phenotypic variability of the disorder. In addition, the presence of a complex *ATXN10* repeat configuration ((ATTCT)<sub>100</sub>(ATTCT)<sub>1</sub>(CTTCT)<sub>38</sub>), despite being within an intermediate range not compatible with SCA10, raises the possibility that non-canonical repeat interruptions could influence repeat stability or exert a subtle modulatory effect on neuronal excitability. While this specific motif architecture has not been previously linked to seizure susceptibility, its unusual structure warrants consideration as a potential modifying factor rather than a primary pathogenic driver. Importantly, this case underscores the need to confirm RP-PCR findings with full repeat sequencing, particularly in admixed populations with large Native American components that have a relatively high frequency of intermediate *ATXN10* alleles.<sup>12</sup> Reliance on RP-PCR alone may lead to misclassifications such as genotype–phenotype correlations or overinterpretation of intermediate expansions.

In our cohort, RP-PCR labeling of “>32 repeats, probable pathogenic” generated diagnostic momentum toward SCA10, despite discordant clinical features and the absence of supportive segregation data. RP-PCR is effective for detecting the presence of a repeat expansion signal, but it does not reliably size large alleles, cannot resolve sequence composition (pure versus interrupted), and can yield results that are over-interpreted when rigid cutoffs are applied. Our cases demonstrate that a threshold such as “>32 repeats = probable pathogenic” is not clinically specific for SCA10 and can inflate false-positive attribution.

The four previous cases from literature suggesting coexistence of SCA10 and an alternative neurogenetic disorder, have larger *ATXN10* expansions; however, clinical phenotypes are mostly concordant with the alternative neurogenetic disorders rather than typical SCA10 phenotypes. Previous studies documenting asymptomatic subjects with pure ATTCT-*ATXN10* expanded alleles, provide supporting evidence to at least consider the possibility that pure *ATXN10* expansion might not be penetrant or cause SCA10 phenotype.

In summary, intermediate *ATXN10* expansions (~100–150 repeats) are unlikely to be pathogenic, and even large, pure ATTCT expansions may be non-pathogenic in some individuals. RP-PCR cutoffs that label >32 repeats as “probable pathogenic” risk overdiagnosis SCA10 and delaying accurate diagnosis. Proper clinical interpretation requires confirmatory sizing and motif-resolution (preferably LRS) and, when clinical-genetic concordance is lacking, additional clinical and genetic testing to identify the underlying cause.

## Declarations

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### Author Roles:

(A) Conceptualization, (B) Investigation, (C) Funding acquisition, (D) Writing - original draft, (E) Methodology, (F) Validation, (G) Visualization, (H) Writing - review & editing, (I) Software, (J) Formal analysis, (K) Project administration, (L) Data curation, (M) Supervision, (N) Resources

**M.C.-O.:** A, B, C, D, E, H, L, M, N  
**A.R.:** B, E, F, G, H, L  
**A.A.:** B, D, E, F, G, H, L, N  
**K.S.:** B, E, G, H, L  
**E.S.-C.:** B, F, G, H, L  
**H.M.S.:** B, E, F, G, H, L  
**C.M.-E.:** B, E, F, H, L  
**C.A.M.T.:** B, E, F, G, H, L  
**M.I.-M.:** A, B, C, F, H, K, L  
**E.D.:** B, E, F, G, H, L  
**A.R.-V.:** B, F, G, H, L  
**W.Q.:** B, E, F, G, H, L  
**B.S.:** A, B, C, D, E, F, H, L, M, N

#### **Disclosures:**

#### **Ethical Compliance Statement:**

This study was approved by the Ethical Committees at *Instituto Nacional de Ciencias Neurológicas*, Lima (014-2025-CIEI-INCN), and *Universidad Científica del Sur* (1067-CIEI-CIENTIFICA-2025). Written informed patient consent was obtained by the authors or the treating physician. We confirm that we have read the Journal's position on issues involved in ethical publication and affirm that this work is consistent with those guidelines.

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## Table

**Table 1 “SCA10 diagnosis” with a coexisting neurodegenerative disorder, summary of published cases.**

|                                        | (Roxburgh RH et al) <sup>6</sup>                                                 | (Kapur SS, Goldman JG) <sup>7</sup>                                            | (Baizabal-Carvallo JF et al.) <sup>8</sup><br>Proband                 | (Baizabal-Carvallo JF et al.) <sup>8</sup><br>Proband's mother                  | Case 1                                                                                     | Case 2                                                                                           | Case 3                       |
|----------------------------------------|----------------------------------------------------------------------------------|--------------------------------------------------------------------------------|-----------------------------------------------------------------------|---------------------------------------------------------------------------------|--------------------------------------------------------------------------------------------|--------------------------------------------------------------------------------------------------|------------------------------|
| <b>Sex, Age at examination (years)</b> | Female, 39                                                                       | Male, 54                                                                       | Male, 39 (proband),                                                   | Female, 63 (proband's mother)                                                   | Female, 34                                                                                 | Male, 56                                                                                         | Male, 44                     |
| <b>Number of cases/families</b>        | 1/1                                                                              | 1/1                                                                            | 2/1                                                                   |                                                                                 | 1/1                                                                                        | 1/1                                                                                              | 1/1                          |
| <b>Age at onset (years)</b>            | mid 30s                                                                          | 43                                                                             | 10 (seizures)                                                         | 20s (seizures), 40s (ataxia)                                                    | 24                                                                                         | 51                                                                                               | 30                           |
| <b>Clinical presentation</b>           | generalized chorea, upper-limb dystonia, gait disturbances, cognitive impairment | gait and limb ataxia, mild dysarthria, slow saccades, oculomotor abnormalities | seizures, cerebellar ataxia, slow saccades, cerebellar atrophy on MRI | seizures, cerebellar ataxia, dysarthria, slow horizontal saccades, hyporeflexia | spasticity, spastic gait, lower-limb spasticity, speech disturbances, cognitive impairment | gait ataxia, dysarthria, autonomic dysfunction, bradykinesia, rigidity, depressive mood, anxiety | seizures, gait spasticity, s |
| <b>ATXN10 repeat expansion</b>         | (ATTCT) <sub>2511</sub>                                                          | (ATTCT) <sub>962</sub>                                                         | (ATTCT) <sub>&gt;850</sub>                                            | (ATTCT) <sub>&gt;850</sub>                                                      | (ATTCT) <sub>92</sub> (ATTCTAT) <sub>8</sub>                                               | (ATTCT) <sub>1127</sub> (ATTTTTTTCT) <sub>1</sub>                                                | (ATTCT) <sub>100</sub>       |
| <b>ATXN10 genotyping technique</b>     | Not reported                                                                     | Not reported                                                                   | PCR-based, Southern Blot                                              | PCR-based, Southern Blot                                                        | PCR/TP-PCR, PCR/RP-PCR, WGS, LRS                                                           | PCR/TP-PCR, PCR/RP-PCR, LRS                                                                      | PCR/TP-PCR                   |
| <b>Coexisting disorder</b>             | HD                                                                               | SCA2                                                                           | SCA2                                                                  | SCA2                                                                            | SPG35                                                                                      | MSA-C                                                                                            | MJD/SCA3                     |
| <b>Diagnostic criteria</b>             | HTT (CAG) <sub>48</sub>                                                          | ATXN2 (CAG) <sub>38</sub>                                                      | ATXN2 (CAG) <sub>exp</sub>                                            | ATXN2 (CAG) <sub>exp</sub>                                                      | FA2H c.461G>A (p.Arg154His)                                                                | 2022 MDS MSA diagnostic criteria (clinically established)                                        | ATXN3 (CTC                   |

**Abbreviations:** PCR, Polymerase Chain Reaction; TP-PCR, Triplet-Primed Polymerase Chain Reaction; PCR/RP-PCR, Repeat-Primed Polymerase Chain Reaction; WGS, Whole-Genome Sequencing; LRS, Long-Read Sequencing

## Videos

Video 1 and 2 are not available with this version.

**Video 1.** Neurological examination of case 1 at age 32 showed markedly impaired spastic gait. The patient remained ambulatory only with walker assistance, taking slow, short steps with reduced foot clearance and instability during walking.

**Video 2.** Neurological examination of case 2 at last follow-up showed gait only with strong support. SARA-based assessment disclosed marked appendicular cerebellar dysfunction, with slightly irregular alternating hand movements predominantly on the left and a marked bilateral intention tremor on nose-finger testing.

Figures

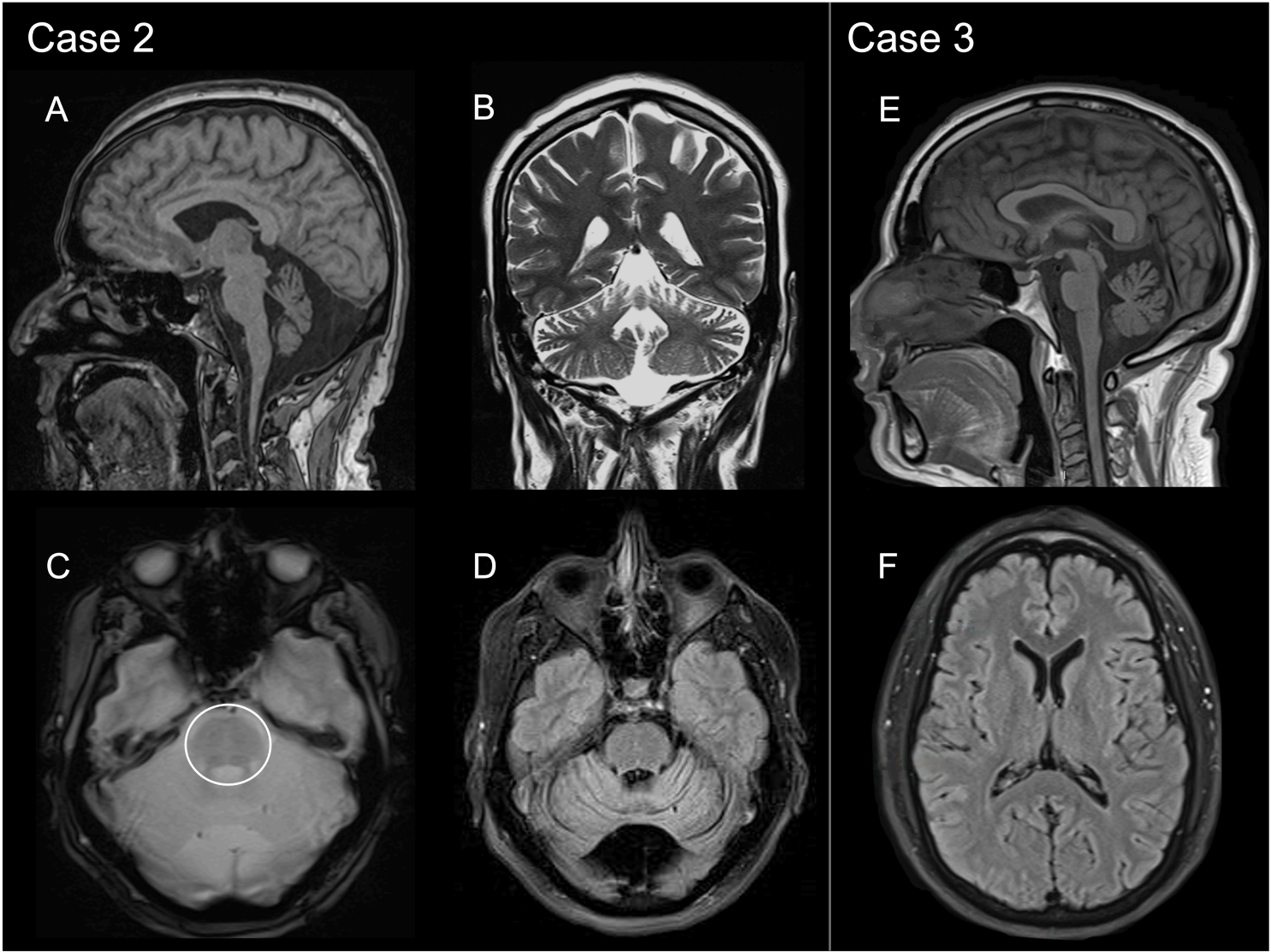


**Figure 1**

Brain MRI of case 2 revealed a retrocerebellar arachnoid cyst with cerebellar atrophy on sagittal T1 (A) and coronal T2 (B). Axial gradient echo highlights the “hot cross bun” sign in the pons (white circle) (C). Axial FLAIR shows arachnoid cyst occupying posterior fossa (D). Case 3 images revealed cerebellar atrophy in sagittal T1 (E), without abnormalities on axial view (F).

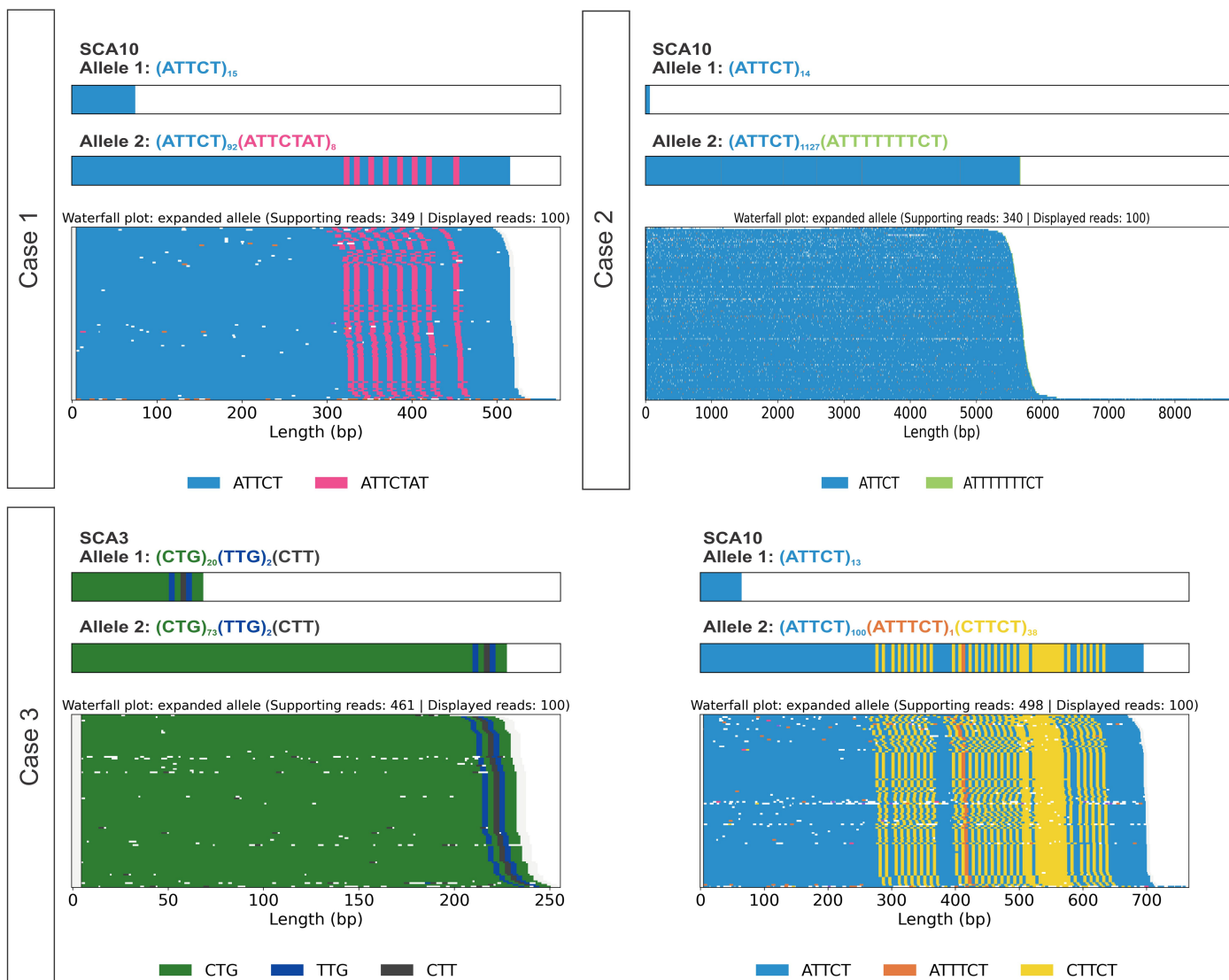


Figure 2

#### SCA10 and SCA3 repeat composition for atypical ataxia cases.

For each case, individual long reads spanning the disease locus are aligned and visualized as waterfall plots (each horizontal line represents one read; x-axis represents position across the repeat tract). The schematic bar above each panel summarizes the repeat tract region and its consensus length in base pairs. Repeat motifs are color-coded as indicated in each panel. Insets show histograms repeat-tract length (bp) for the displayed expanded alleles. Genotype calls are listed below each panel: **(A)** Case 1 shows an interrupted expansion with mixed repeat motifs (ATTCT)<sub>92</sub>(ATTCTAT)<sub>8</sub>; (ATTCT)<sub>100</sub>(ATTCT)<sub>1</sub>(CTTCT)<sub>38</sub>. **(B)** Case 2 (SCA10) shows a large, predominantly pure ATTCT expansion (14 and 1127 ATTCT repeats); **(C)** Case 3 shows CTG repeat alleles of 11 and 20 in SCA3 and an interrupted expansion with an ATTCTAT component.

## Supplementary Files

This is a list of supplementary files associated with this preprint. Click to download.

- [Supp.InformationCSSCA10MDCP.docx](#)