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Case Report

Laminectomy as treatment for abrupt neurological decline in acrodysostosis: A case report

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ABSTRACT

Background: Acrodysostosis (ACRO) is a rare disorder of peripheral bone development which can be either sporadic or inherited with mutations in the *PRKAR1A* or *PDE4D* genes. The resulting phenotypical characteristics are variable and overlap with other dysostosis conditions, making diagnosis difficult without genotyping. Vertebral malformations have been reported with ACRO resulting in slowly progressive spinal cord compression leading to radiculopathy or myelopathy.

Case Description: A 19-year-old female diagnosed with ACRO presented with progressively worsening lower extremity paraparesis, sensory loss, and urinary retention; she was wheelchair-bound. A magnetic resonance imaging showed cord signal change at the T2/T3 levels with accompanying diffuse cord edema between T6-T8. Six months following a T2/T3 and T6/T7 laminectomy, the patient's symptoms improved, but she still required a wheelchair.

Conclusion: Patients with ACRO should be regularly monitored for cord compression to allow for early surgical decompression to prevent long-term, devasting neurological compromise.

Keywords: Acrodysostosis, Laminectomy, Neurosurgery

INTRODUCTION

Acrodysostosis (ACRO) is a disorder in peripheral bone development initially described in 1968 by Maroteaux.[4] It is a member of the dysostoses, characterized by bone malformations attributed to blastogenesis abnormalities.[6] It is a rare pathology, with <70 cases reported in the literature.[9] Signature characteristics of the disease include: brachydactyly affecting all metacarpals, diffuse peripheral dysostosis, short stature, skull hypoplasia, maxillonasal hypoplasia, and shortened interpedicular distance due to premature closure of vertebral epiphyses, which contributes to spinal stenosis.[3] Case reports of familial inheritance identified two genes, *PRKAR1A* and *PDE4D*, which are responsible for ACRO.[7] Here, we discuss a patient with ACRO who presented with thoracic myelopathy requiring T2/3 and T6-8 decompressive laminectomies.

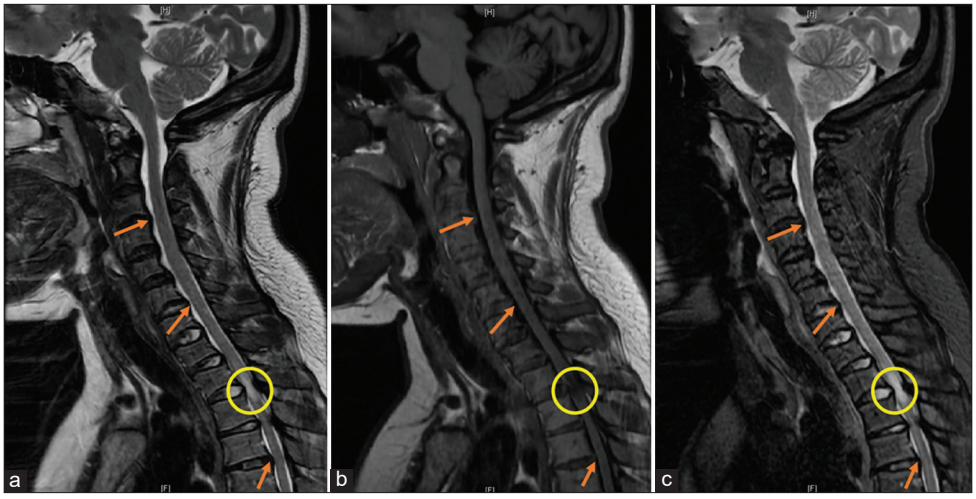

Figure 1: (a) Sagittal T2-weighted magnetic resonance imaging (MRI) of the cervical spine showing a hyperintensity and buckling of ligamentum flavum (circle) at T2-T3 and canal stenosis (arrows) at multiple levels. (b) Sagittal T1-weighted MRI showing hypointensity and buckling of the ligamentum flavum (circle) at T2-T3 and canal stenosis (arrows) at multiple levels. (c) Sagittal short T1-weighted inversion recovery (STIR) MRI showing hyperintensity and buckling of the ligamentum flavum (circle) at T2-T3 consistent with cord edema and canal stenosis (arrow) at multiple levels.

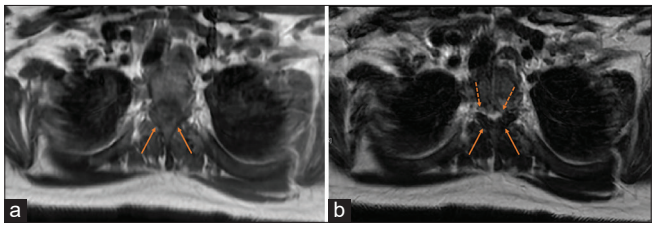

Figure 2: (a) Axial T1-weighted FLAIR MRI at T2/T3 level showing thickening/hypertrophy of ligamentum flavum (solid arrows) and decreased spinal canal diameter. (b) Axial T2-weighted MRI at the T2/T3 level showing thickening/hypertrophy of ligamentum flavum (solid arrows) and cord signal change (dashed arrows) moreprominent on the left than the right.

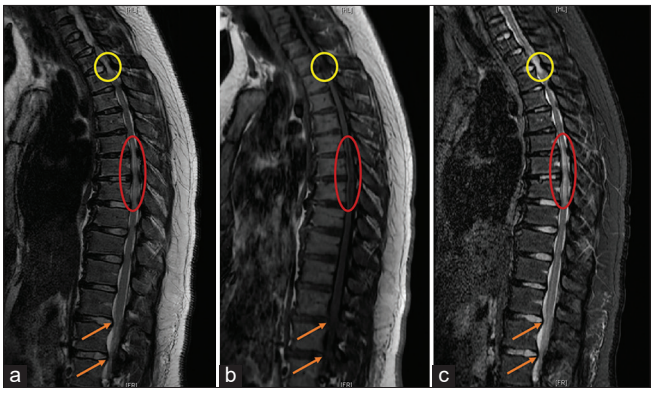

Figure 3: (a) Sagittal T2-weighted MRI showing a hyperintensity of the cord and buckling of ligamentum flavum (circle) at T2-T3, diffuse hyperintensity (oval) from T6-T8, and lumbar stenosis (arrows). (b) Sagittal T1-weighted MRI showing hypointensity of the cord and buckling of the ligamentum flavum (circle) at T2-T3, diffuse hypointensity (oval) from T6-T8, and lumbar stenosis (arrows). (c) Sagittal short T1-weighted inversion recovery (STIR) MRI showing hyperintensity and buckling of the ligamentum flavum (circle) at T2-T3 consistent with cord edema, diffuse hyperintensity (oval) from T6-T8, and lumbar stenosis (arrow).

CASE DESCRIPTION

A 19-year-old female with a history of ACRO presented with a 3-month complaint of progressive paraparesis leading to paraplegia, a loss of pinprick sensation at the T4 level, and urinary retention. Magnetic resonance imaging showed central canal stenosis that was more prominent on the left, cord signal change at the T2/T3 level [Figures 1 and 2], and diffuse cord edema from the T6-T8 levels [Figures 3 and 4]. She underwent T2/T3 and T6/T7 level laminectomies and was discharged on postoperative day 4. Despite surgical decompression, at 3-month follow-up, she still had significant residual paraparesis, urinary retention, and required a wheelchair.

DISCUSSION

We found only five instances of ACRO that were myelopathic and attributable to MR-documented stenosis. [Table 1] Three out of five cases underwent surgical treatment, with the other two undergoing medical management.[1-3,5,8] One case similar to ours involved a 22-year-old man with mild left lower extremity motor paresis (4/5) and left T4–5 sensory level. One month following a T3–5 laminectomy, the patient could ambulate, and the postoperative MR showed adequate spinal cord decompression.[3]

Table 1: Table showing the characteristics of acrodysostosis patients with neurological deficits reported in the literature, including our case, listing the patient's presenting signs/symptoms, imaging findings, treatment approach, and length of follow-up.

Author (date)	Age (years)	Signs/Symptoms	Imaging	Treatment	Outcome	Follow-up
Current case	19	Paraparesis that had progressed to paraplegia, progressive sensory loss below the level of T4, developed urinary retention a few days prior to surgery	Left predominant central canal stenosis with cord signal change at T2/T3 level, diffuse cord edema from the T6–8 levels	T2/T3 and T6/T7 laminectomy	paraplegia and urinary retention remained but no longer progressing, subjective improved hip control, improved sensation below the T4/T5 level	3 months
Martín *et al*. (2021)	8	dyspnea with moderate physical exertion (Class II–III dyspnea) moderate non-reversible obstructive ventilatory deficit with marked air trapping on plethysmography	None	corticosteroids, azithryomycin, monetlukast, combined high-dose inhaled treatment and respiratory physiotherapy	spirometry showed slow progressive improvement, stable on low-dose combination inhaled therapy with no dyspnea on exertion	4 years
Jalabert *et al*. (2018)	48	bilateral lower extremity radicular pain in the S1 dermatome no sexual, bowel, or bladder dysfunction	mild degenerative disk disease, "fish-shaped" vertebral bodies, facet joint osteoarthritis at the lumbar levels on T2-weighted MRI	flexion-based exercise therapy, endurance training, and spinal injections with epidural and intradural corticosteroids	sustained symptomatic relief	NR
Lahoud *et al*. (2014)	22	progressive left lower extremity paresthesia and proximal weakness requiring crutches 4/5 proximal lower limb monoparesis left babinski sign no bowel/bladder dysfunction somatosensory evoked potentials showed alteration of signal conduction from T1-T112 in the lower extremity	extensive cervical and thoracic spinal canal stenosis, hypertrophied ligamentum flavum at the T4–5 level, increased signal in spinal cord at T4-T5 level	IV corticosteroids followed by T3–5 laminectomy	sensory-motor recovery seen on POD 4, regain mobility without the need for crutches and complete recovery of motor and sensory function at last follow-up	1 month
Sezer *et al*. (2009)	43	low back pain, difficulty walking, and weakness of arms and legs of several years in duration became progressive over a year, presented to neurosurgery clinic after failed trail of anti-rheumatic drug treatment and becoming wheelchair-bound	MRI showed diffuse spinal stenosis, myelopathy and disc pathology at multiple levels of cervical and thoracolumbar regions	initial surgery was C5-C median corpectomy, C4 corpus wedge resection, and C4–7 anterior stabilization and graft followed by laminectomy and foraminotomy at multiple thoracolumbar levels 3 months later	almost complete motor recovery in the upper extremity and partial recovery in the lower extremity postoperatively at last follow up, strength was 4/5 in the upper extremity and after intensive rehabilitation program was able to walk with a walker and orthopedic shoes without pain or spasticity	3 months
Hamanishi *et al*. (1993)	35	paresthesia on lateral aspect of the right upper arm, forearm, and hand paresthesia and pain on lateral aspect of bilateral thighs, legs, and dorsum of feet no bowel or bladder dysfunction	narrowing at C7-T1 level, diffuse stenosis of lumbar spine with compression at L2-L5 levels	patient deferred open laminectomy	temporary relief of sciatica and intermittent claudicatoin by continuous pelvic traction, NSAIDs and prostaglandin-E1	NR

Table 2: Table of included references as well as summaries of the articles.

Title	Author	Journal	Year of publication	Summary
Acrodysostosis associated with spinal canal stenosis	Hamanishi C., Nagata Y., Nagao Y., Sohen S., Tanaka S	Spine	1993	Case report of an adult female patient with acrodysostosis with lumbar spinal canal stenosis. Had right arm numbness, intermittent neurogenic claudication with numbness on both legs and pain/weakness in the left leg.
Radicular claudication revealing possible acrodysostosis: A case report	Jalabert M., Rannou F., Nguyen C	Annals of Physical and Rehabilitation Medicine	2018	Letter to the editor of adult male with a late diagnosis of acrodysotosis that was diagnosed on presentation due to radicular pain. Treated with home-based flexion-based exercise therapy, endurance training, and spinal injections that led to adequate relief for the patient.
Acrodysostosis and spinal canal involvement	Lahoud GA., Chalouhi N., Jabbour P	World Neurosurgery	2014	Case report of an adult male with acrodysostosis and spinal cord compression at T4/T5 level causing lower limb monoparesis, left T6 sensation decrease, left hyperreflexia and left Babinski sign. Underwent T3–5 laminectomy with near complete recovery. First report of histological description in literature of bony abnormalities in acrodysostosis
Acrodysostosis	Maroteaux P., Malamut G	La Presse médicale	1968	First description of acrodysostosis in the literature
Radiological diagnosis of the constitutional disorders of bone. As easy as A, B, C?	Offiah AC., Hall CM	Pediatric Radiology	2003	Gives a radiological description of several constitutional disorders of bone including acrodysostosis
Gαs-Protein Kinase A (PKA) Pathway Signalopathies: The Emerging Genetic Landscape and Therapeutic Potential of Human Diseases Driven by Aberrant Gαs-PKA Signaling	Ramms DJ., Raimondi F., Arang N., Herberg FW., Taylor SS., Gutkind JS	Pharmacological Reviews	2021	Review article of the different pathologies involving the Gαs–Protein Kinase A Pathway and the genes involved in the diseases. Acrodysostosis and the genes involved are discussed specifically in parathyroid hormone intracellular signaling.
Adult case of acrodysostosis with severe neurologic involvement	Sezer N., Sutbeyaz ST., Koseoglu F., Aras M., Akin C	Journal of Back and Musculoskeletal	2009	Case report of an adult female patient with longstanding history of neck and back pain with weakness underwent initial cervical corpectomy at C5–6 level followed by laminectomy and foraminotomy at several thoracolumbar levels with almost complete reversal of symptoms at last follow-up
Respiratory impairment in a patient with acrodysostosis: A rare association of an uncommon pathology	Martín Sierrasesúmaga P., Zubiri Berrade .S, Guindulain Chueca MJ., Viguria N., Moreno-Galarraga L	Archivos Argentinos de Pediatria	2021	Case report of a female pediatric patient with genetically confirmed acrodysostosis with progressive lung function deterioration. It was the first reported case of acrodysostosis associated with respiratory dysfunction, had improvement after rigorous medical management.
Acrodysostosis	Silve C., Clauser E., Linglart A	Hormone and Metabolic Research	2012	Review article of acrodysostosis of clinical, radiological, and metabolic features of acrodysostosis followed by the recent developments in understanding of the pathogenesis of acrodysostosis and the similarities or differences between this and other diseases that are related to the parathyroid hormone signaling pathway
The association between ankylosing spondylitis and the risk of any, hip, or vertebral fracture: A meta-analysis	Zhang M., Li M., Wang GS., Tao JH., Chen Z., Ma Y., and Li XP	Medicine (Baltimore)	2017	A meta-analysis on the association between ankylosing spondylitis and the risk of any, hip, or vertebral fracture. They found that ankylosing spondylitis was strongly associated with vertebral fracture but not any fracture

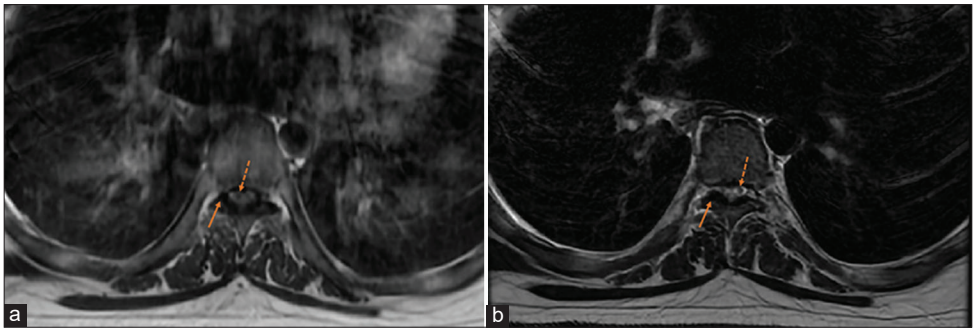

Figure 4: (a) Axial T1-weighted FLAIR MRI at T6/T7 level showing thickening/hypertrophy of ligamentum flavum (solid arrows) and slight cord change (dashed arrows). (b) Axial T2-weighted MRI at T6/T7 level showing thickening/hypertrophy of ligamentum flavum (solid arrows) and slight cord change (dashed arrows).

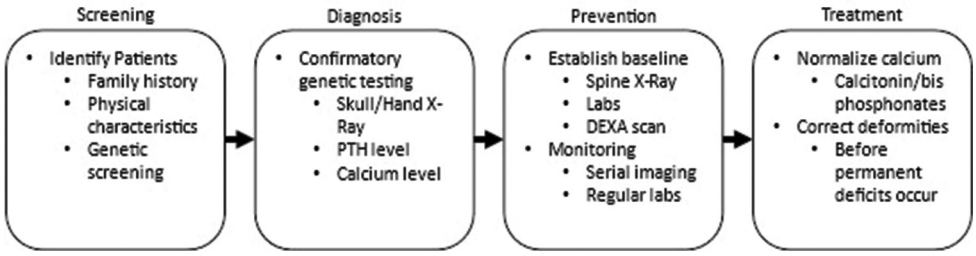

Figure 5: Our proposed algorithm for screening and diagnosing acrodysostosis and preventing complications before they arise or treating them before severe neurological deficits set in.

Acrodysotosis: Differential diagnostic considerations

The differential diagnostic considerations for ACRO include idiopathic hypercalcemia, pseudohypoparathyroidism, and ankylosing spondylitis (AS).[9,10] More detailed discussions of these differentials in addition to other characteristics of ACRO is outside the scope of this paper , but can be found in the summarized literature [Table 2]. While most ACRO cases are sporadic, there is also a hereditary component.[7] One may use a similar screening and monitoring algorithm for ACRO as that applied to AS, a component of which may include genetic testing [Figure 5].

CONCLUSION

Patients with ACRO, either sporadic or genetic in origin, should be routinely monitored for the onset of radiculopathy and/or myelopathy reflecting progressive cord compromise due to stenosis, which may or may not warrant surgical spinal decompression.

Declaration of patient consent

Patient's consent not required as patient's identity is not disclosed or compromised.

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Nil.

Conflicts of interest

There are no conflicts of interest.

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