HORIZONTAL GAZE PALSY WITH PROGRESSIVE SCOLIOSIS SYNDROME (HGPPS): CASE PRESENTATION AND BRIEF REVIEW OF THE CURRENT LITERATURE

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PURPOSE

To present a case of Horizontal Gaze Palsy with Progressive Scoliosis Syndrome (HGPPS) and conduct a brief review of the current literature

MATERIALS-METHODS

Retrospective review of the patient's notes

RESULTS

A 3-year-old male child was referred to the paediatric ophthalmology clinic of Leicester Royal Infirmary due to persistent squint since he was 6 months old. He also encountered significant problems with his walking ability and body posturing.

Skeletal examination revealed scoliosis and inability to stand upright without support [1]. Binocular visual acuity fluctuated between 6/12 and 6/9 on the Snellen chart [1]. Orthoptic evaluation revealed bilateral horizontal gaze palsy with abduction deficit, esotropia of 35 diopters, and pendular nystagmus [1]. Vertical eye movements, convergence and pupillary reflexes were normal [1]. MRI brain revealed bilateral hypoplastic sixth nerve nuclei with brainstem hypoplasia, the presence of a deep midline pontine cleft, and a butterfly shaped medulla [1]. MRI brain figures are shown in slide 2 of this poster.

The child was referred to the genetic department and mutation in the ROBO-3 gene was confirmed [1]. These findings were compatible with the diagnosis of HGPPS. After 5 years of follow-up, the condition has remained stable.

FOOTNOTE-DECLARATION
THIS ELECTRONIC POSTER IS A BRIEF SUMMARY OF THE FOLLOWING PUBLISHED MANUSCRIPT: Tyradellis, Straton MD; Tsokolas, Georgios MSc; Pillai, Swati MD. Horizontal gaze palsy with progressive scoliosis syndrome in a paediatric patient: A case report. Medicine Case Reports and Study Protocols 2(11):p e0164, November 2021. | DOI: 10.1097/MD9.0000000000000164 [1]

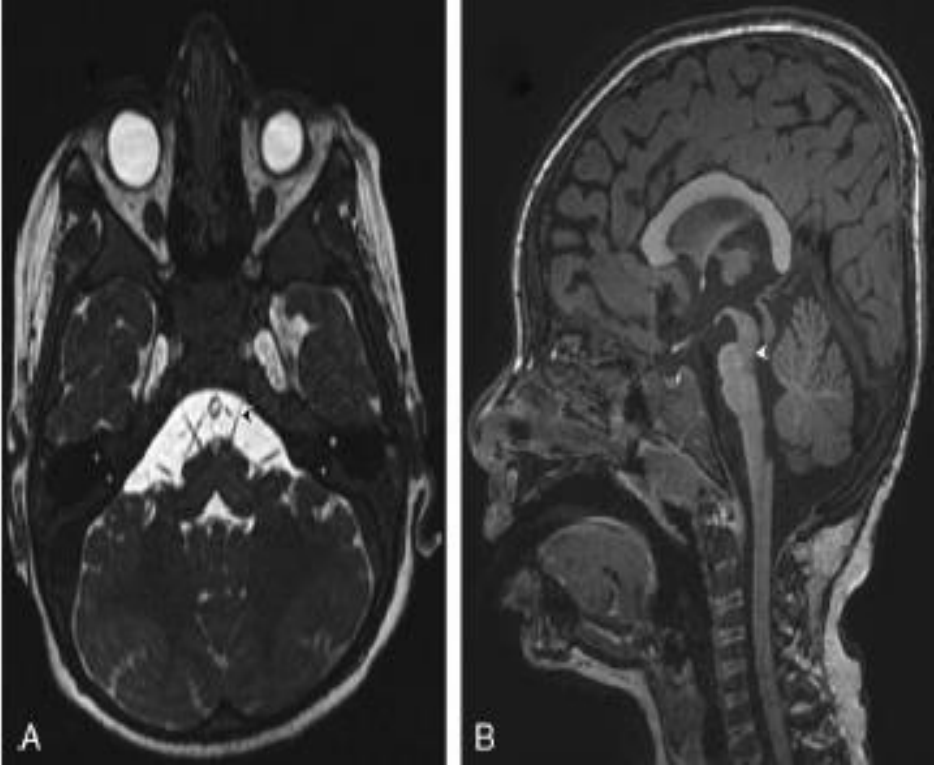

Figure 1. From left to right: A) Axial FIESTA Sequence showing cisternal portion of the abducent nerve (Small Black Arrow) [1] B) Sagittal T1 Sequence showing hypoplastic brainstem with depression of the fourth ventricle (Small White Arrow) [1] (MRI Brain image from Tyradellis et al. [1])

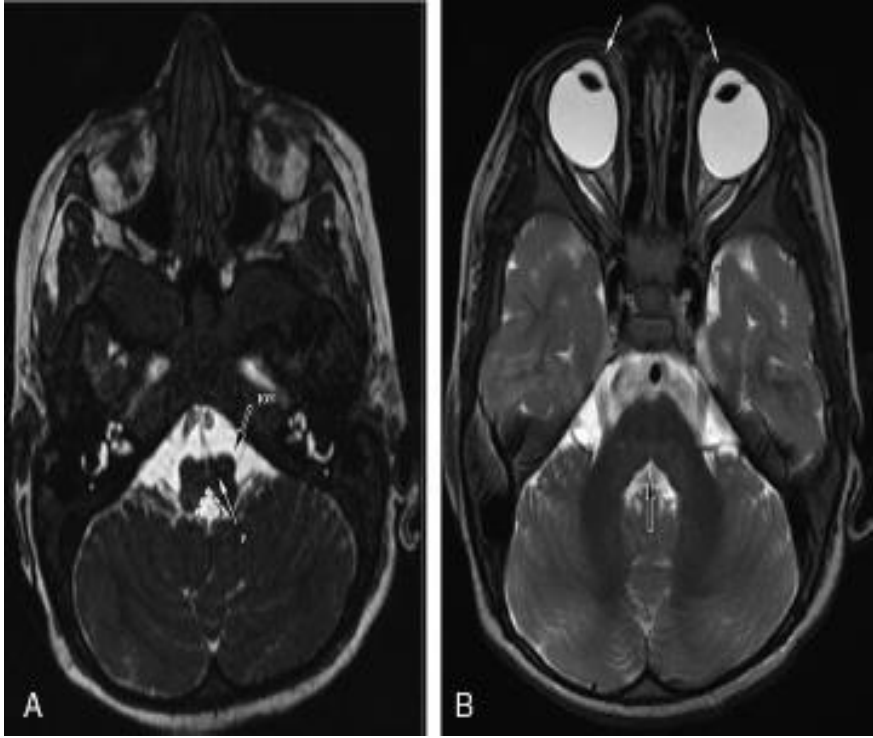

Figure 2. From left to right: A) A) Axial FIESTA Sequence showing butterfly configuration of the medulla with midline cleft. Also of note is the prominent Inferior Olivary Nucleus (ION) (Black Arrow) with respect to Pyramid (P). Furthermore, the white dashed arrows show tenting of the fourth ventricle [1]. B) Axial T2 showing bilateral convergent strabismus. In addition, there is dorsal pontine cleft and absent facial colliculus (Black Arrow) [1] (MRI Brain image from Tyradellis et al. [1])

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DISCUSSION

HGPPS is a rare autosomal recessive genetic condition affecting the central nervous system, the eyes, and the musculoskeletal system [2]. Based on our literature review, to date, roughly 100 patients with this rare syndrome and 55 ROBO-3 gene mutations have been reported [2, 3].

The age of onset of symptoms may vary from 2 months up to 60 years, but usually start in early childhood [1, 2]. Musculoskeletal symptoms include scoliosis of the spine and difficulties with posturing [1, 2]. Mental retardation and global developmental delay may also ensue. Ophthalmological symptoms may include convergent squint, pendular nystagmus and horizontal gaze palsy [1, 2]. Vertical and convergence eye movements are usually preserved [1, 2]. Usually, the same is the case regarding the lid position, pupillary reflexes and fundus examination as well [1, 2]. Brain neuroimaging reveals striking radiological findings, including brainstem hypoplasia with an anterior and dorsal pontine cleft and a butterfly shaped medulla and absence of the normal promontories of the facial colliculi [1, 2].

To date, there is no definitive treatment of this condition [1, 2]. Convergent squint can be corrected with prisms or surgery [1, 2]. Spectacles are used to correct any significant refractive error [1, 2]. Patching or atropine drops can be used for the treatment of amblyopia [1, 2].

CONCLUSION

This rare syndrome is a systemic condition that requires a multidisciplinary team to treat the patients with a holistic approach [2]. It is recommended that various allied healthcare professionals should be involved in the management of HGPPS patients, including paediatric neurologists, orthopaedic surgeons, ophthalmologists, speech and language therapists, physiotherapists, orthoptists, psychologists and social services [1, 2]. This is of paramount importance as this will allow a better and more all-around clinical care [1, 2]. Finally, it will facilitate the provision of stronger emotional support to HGPPS patients and their carers and will enable them to deal a lot better with the anxiety and stress caused by the challenging day-to-day living with this incurable condition [1, 2].

REFERENCES

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3) **Yi S, Qin Z, Zhou X et al. Early onset horizontal gaze palsy and progressive scoliosis due to noncanonical splicing-site variant and a missense variant in the ROBO3 gene. Mol Genet Genomic Med. 2023 ;11(9): e2215**

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