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Cochlear Implantation in Radiologically Compatible Chudley-Mccullough Syndrome: A Familial Case Series of Three Sisters

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Abstract | **Case Report**

Chudley-McCullough syndrome is a rare autosomal recessive disorder characterized by sensorineural hearing loss associated with distinctive brain malformations on magnetic resonance imaging. Published data on cochlear implantation in this condition remain limited. We report three sisters aged 18, 15, and 3 years with prelingual bilateral profound sensorineural hearing loss and a neuroradiologic pattern compatible with Chudley-McCullough syndrome, including corpus callosum abnormalities, parasagittal frontal polymicrogyria, heterotopia, and arachnoid cysts. Inner-ear structures and cochlear nerves were preserved in all three patients. All underwent unilateral cochlear implantation through mastoidectomy and posterior tympanotomy, with complete electrode insertion and no immediate postoperative complication. Follow-up was short, with postoperative assessments available at approximately 1 month in the eldest sister and approximately 3 months in the two younger sisters. Early auditory behavioral improvement was observed, particularly in the youngest patient, although outcome interpretation remains limited by the retrospective estimation of Categories of Auditory Performance and short follow-up. This familial series suggests that, in patients with profound hearing loss and imaging findings radiologically compatible with Chudley-McCullough syndrome, cochlear implantation may be considered when cochlear nerves are present and inner-ear anatomy is favorable. Molecular confirmation remains desirable for diagnostic consolidation and genetic counseling, but auditory rehabilitation should not necessarily be delayed solely for genetic testing when the clinical and radiological context is strongly suggestive.

Keywords: Chudley-McCullough syndrome; sensorineural hearing loss; cochlear implantation; polymicrogyria; heterotopia; corpus callosum agenesis.

INTRODUCTION

Congenital hearing loss is a common clinical condition, and genetic causes account for a substantial proportion of permanent childhood sensorineural hearing loss [1]. Syndromic forms may be suggested by associated neurological, craniofacial, ocular, renal, or radiological features. Among these rare entities, Chudley-McCullough syndrome is characterized by sensorineural hearing loss associated with a recognizable pattern of brain malformations [2-4].

The reported neuroradiological spectrum includes partial or complete agenesis of the corpus callosum, medial or parasagittal frontal polymicrogyria, periventricular or pericallosal heterotopia, arachnoid cysts, cerebellar dysgenesis, and ventriculomegaly [2-4]. Pathogenic variants in GPSM2 have been identified in many affected families, confirming the genetic basis of the syndrome [3]. A striking feature of the condition is the possible dissociation between extensive structural brain abnormalities and a relatively unremarkable neurological examination [2,4].

Cochlear implantation is a major rehabilitative option in profound sensorineural hearing loss when the cochlear nerve is present and implant anatomy is favorable. However, published experience with cochlear implantation in Chudley-McCullough syndrome remains limited to isolated reports and small series [5,6]. We report a familial case series of three sisters with prelingual bilateral profound sensorineural hearing loss and a magnetic resonance imaging pattern radiologically compatible with Chudley-McCullough syndrome, focusing on implant candidacy, surgical decision-making, and early postoperative auditory behavior.

CASE REPORT

Clinical, audiological, radiological, surgical, and early postoperative data were retrospectively reviewed in three sisters managed in the Department of Otorhinolaryngology and Head and Neck Surgery, Hopital des Specialites, Ibn Sina University Hospital Center, Rabat, Morocco. Written informed consent for publication of anonymized clinical and imaging data was obtained from the adult patient and from the legal guardians of the minor patients.

The three sisters were aged 18, 15, and 3 years at the time of assessment. All had prelingual bilateral profound sensorineural hearing loss documented by auditory evoked potentials. The parents were non-consanguineous. Pregnancies had been poorly monitored, but deliveries were vaginal and reportedly uncomplicated. There was no history of neonatal distress, significant jaundice, exposure to ototoxic drugs, or severe neonatal infection. Vaccinations were up to date according to the national immunization program.

Otoscopic examination was normal in all three patients. The two older sisters had not received formal schooling and communicated primarily through sign language. No motor deficit or seizure history was reported. Neurological examination was unremarkable in all three patients, despite the presence of structural brain abnormalities on imaging.

High-resolution magnetic resonance imaging of the temporal bones and internal auditory canals showed preserved cochleovestibular anatomy, patent internal auditory canals, and visible cochlear nerves in all three sisters, supporting anatomical candidacy for cochlear implantation. Brain magnetic resonance imaging in the eldest sister showed a characteristic pattern combining bilateral symmetric pericallosal laminar heterotopia, bilateral parasagittal frontal polymicrogyria, and partial agenesis of the corpus callosum (Figure 1A-D). Temporal bone computed tomography also identified a contralateral high-riding dehiscent jugular bulb with intratympanic protrusion, closely related to the round window region, which influenced side selection for cochlear implantation (Figure 1E).

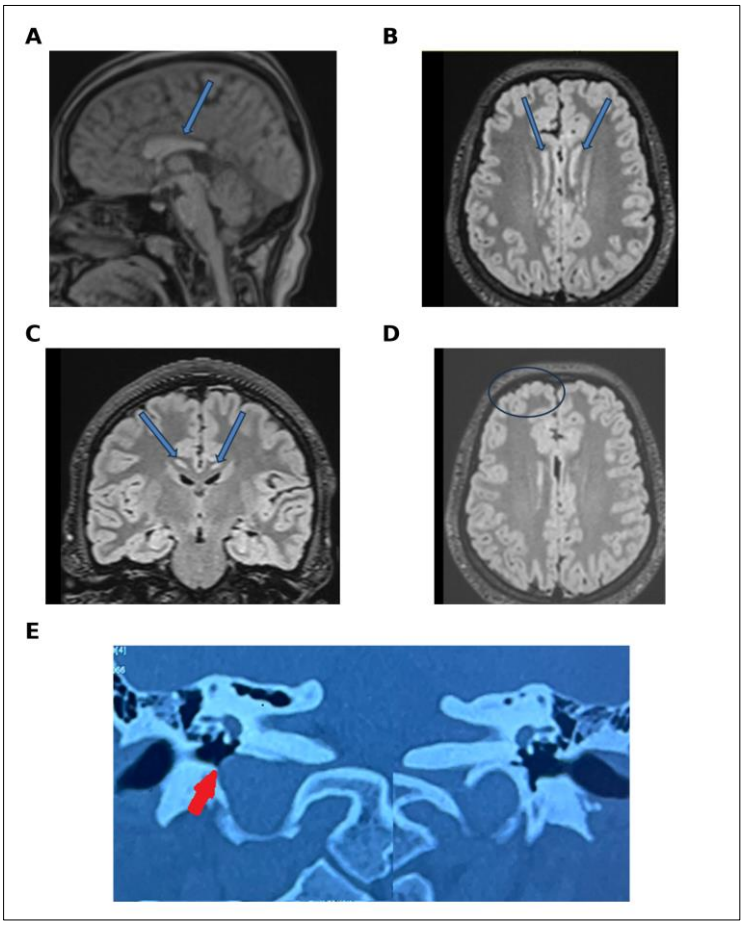

Figure 1. Brain MRI and temporal bone CT in Patient 1. A. Sagittal T1-weighted image showing partial agenesis of the corpus callosum. B. Axial 3D FLAIR image demonstrating bilateral symmetric pericallosal laminar gray matter heterotopia. C. Coronal 3D FLAIR image confirming bilateral pericallosal laminar heterotopia. D. Axial 3D FLAIR image showing bilateral parasagittal frontal polymicrogyria. E. Coronal temporal bone CT image showing a high-riding dehiscent jugular bulb with intratympanic protrusion, closely related to the round window region, which influenced side selection for cochlear implantation.

In the second patient, brain magnetic resonance imaging demonstrated a well-circumscribed quadrigeminal cistern arachnoid cyst measuring approximately 19 x 24 x 27 mm and extending toward the falx cerebri, associated with partial agenesis of the corpus callosum (Figure 2A-B). No major inner-ear malformation was observed, and the cochlear nerves were visualized.

In the youngest patient, brain magnetic resonance imaging showed a large left-sided interhemispheric arachnoid cyst measuring approximately 81 x 64 x 92 mm, with mass effect and ventricular distortion. Associated malformations included partial agenesis of the splenium, absence of the frontal horns, bilateral parasagittal frontal polymicrogyria, band heterotopia along the corpus callosum, and cerebellar dysgenesis, as described in the radiology report (Figure 2C). Inner-ear anatomy was globally preserved, and the cochlear nerves were visible.

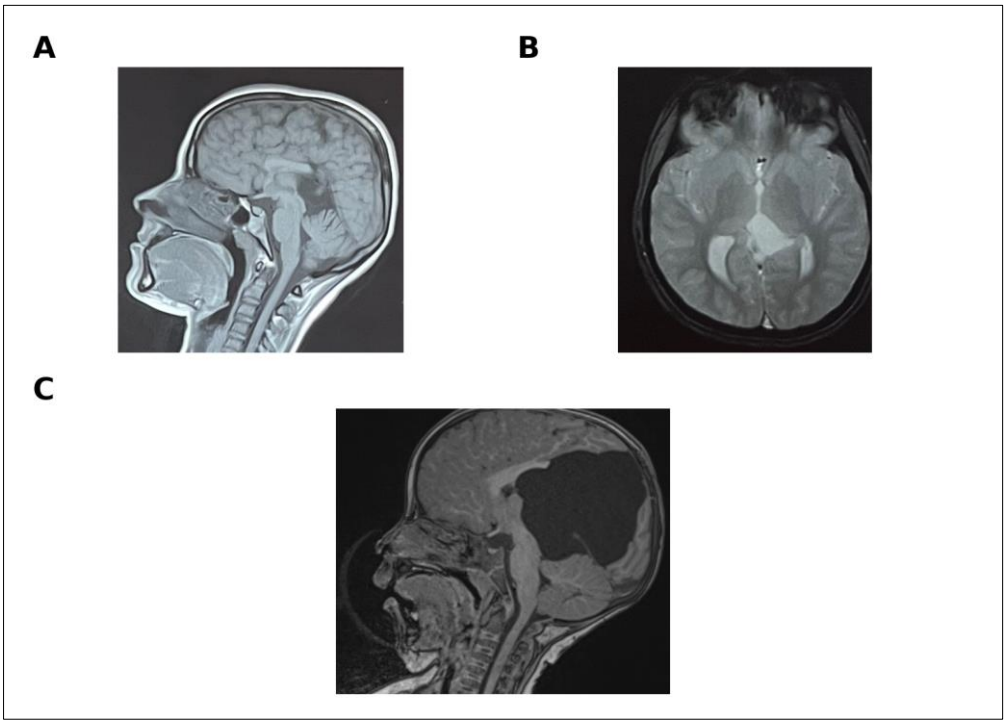

Figure 2. Brain MRI in Patients 2 and 3. A. Sagittal T1-weighted image in patient 2 showing partial agenesis of the corpus callosum. B. Axial T2-weighted image in patient 2 demonstrating a well-circumscribed quadrigeminal cistern arachnoid cyst extending toward the falx cerebri. C. Sagittal T1-weighted image in patient 3 showing a large left-sided interhemispheric arachnoid cyst with marked mass effect; associated cortical and callosal malformations are detailed in the main text and radiology report.

Abbreviations: CAP = Categories of Auditory Performance; CT = computed tomography; IAC = internal auditory canal; MRI = magnetic resonance imaging.

Taken together, the familial clustering of profound congenital hearing loss, the presence of callosal abnormalities, parasagittal frontal polymicrogyria, heterotopia, and arachnoid cysts, and the absence of another unifying diagnosis supported the diagnosis of a radiologically compatible Chudley-McCullough syndrome. Genetic testing was not available and was therefore not performed.

All three patients underwent unilateral cochlear implantation in 2025 by the same surgeon. The procedure consisted of mastoidectomy with posterior tympanotomy in each case. Electrode insertion was complete in all three patients, and no perioperative or immediate postoperative complication was recorded. Device activation was performed approximately 1 month after surgery, followed by fitting sessions according to the department's usual protocol.

Postoperative follow-up remained limited. Assessment was available at approximately 1 month in the eldest sister, who had undergone surgery more recently, and at approximately 3 months in the two younger sisters. Early auditory behavior was assessed using estimated Categories of Auditory Performance, based on clinical observation and caregiver reports. The youngest patient showed the most favorable early auditory trajectory, whereas improvement in the two older sisters was more limited and should be interpreted cautiously because of late implantation, short follow-up, and retrospective outcome assessment.

Table 1: Clinico-radiological summary

Patient	Clinical profile	Key brain MRI findings	IAC MRI / inner-ear findings
1 18 years	Prelingual profound bilateral sensorineural hearing loss; sign language; normal otoscopy and neurological examination.	Bilateral parasagittal frontal polymicrogyria, bilateral pericallosal laminar heterotopia, and partial agenesis of the corpus callosum.	Patent IACs; cochlear nerves visualized; preserved cochleovestibular anatomy.
2 15 years	Prelingual profound bilateral sensorineural hearing loss; sign language; normal otoscopy and neurological examination.	Quadrigeminal arachnoid cyst, approximately 19 x 24 x 27 mm, associated with partial agenesis of the corpus callosum.	No major inner-ear malformation; cochlear nerves visualized.
3 3 years	Prelingual profound bilateral sensorineural hearing loss; normal otoscopy and neurological examination.	Large left interhemispheric arachnoid cyst, approximately 81 x 64 x 92 mm, with mass effect; partial agenesis of the splenium; absent frontal horns; bilateral parasagittal frontal polymicrogyria; band heterotopia; cerebellar dysgenesis.	Cochlear nerves visualized; inner-ear structures globally preserved.

Note: IAC = internal auditory canal; MRI = magnetic resonance imaging. MRI findings are summarized from the available radiology reports.

Table 2: Surgical data and estimated early auditory outcomes

Patient	Implanted side	Specific surgical factor	Follow-up available	Preoperative CAP	Postoperative CAP
1 18 years	Left	Contralateral high-riding dehiscent jugular bulb with intratympanic protrusion.	Approx. 1 month	0-1	1-2 Early estimate
2 15 years	Right	No specific intraoperative difficulty.	Approx. 3 months	0-1	2-3
3 3 years	Right	No specific intraoperative difficulty.	Approx. 3 months	0-1	3-4

Note: CAP = Categories of Auditory Performance. CAP categories were estimated retrospectively from available clinical observations and caregiver reports. Ranges indicate uncertainty related to short follow-up and non-standardized retrospective assessment.

DISCUSSION

This familial case series describes three sisters with prelingual bilateral profound sensorineural hearing loss and a neuroradiological pattern strongly suggestive of Chudley-McCullough syndrome. The main practical point is that extensive cerebral malformations did not preclude cochlear implantation because inner-ear anatomy was preserved and the cochlear nerves were visible on internal auditory canal magnetic resonance imaging.

The diagnostic wording used in this report is deliberately cautious. Chudley-McCullough syndrome is classically associated with severe-to-profound sensorineural hearing loss and a recognizable constellation of brain abnormalities, including callosal agenesis or dysgenesis, medial or parasagittal frontal polymicrogyria, heterotopia, arachnoid cysts, cerebellar dysgenesis, and ventriculomegaly [2-4]. The three sisters displayed several of these features, particularly the eldest and youngest patients. However, because genetic testing was not available, the phrase radiologically compatible Chudley-McCullough syndrome is more accurate than a definitive molecular diagnosis.

Genetic confirmation remains important for diagnostic consolidation, recurrence-risk counseling, and family screening. Pathogenic GPSM2 variants have been identified in many families with Chudley-McCullough syndrome [3]. Nevertheless, in a child with profound hearing loss and a highly suggestive imaging pattern, the immediate otological priority is to determine whether auditory rehabilitation is anatomically feasible. In this setting, documentation of cochlear nerve presence and preserved cochleovestibular anatomy is central to implant candidacy [5,6].

Published data on cochlear implantation in Chudley-McCullough syndrome remain limited, but available reports suggest that auditory benefit can be obtained when the cochlear nerve is present [5,6]. The syndrome is not primarily defined by inner-ear malformations. Therefore, associated cerebral malformations should not automatically be considered a contraindication to cochlear implantation. Instead, they

should inform counseling, expectations, and follow-up planning.

The early postoperative findings in the present series should be interpreted with caution. Follow-up was short, and Categories of Auditory Performance were estimated retrospectively from clinical observations and caregiver reports. These findings therefore reflect early auditory behavioral responses rather than definitive auditory-language outcomes. The more favorable early trajectory in the youngest patient is consistent with established principles of auditory neuroplasticity and the influence of age at implantation on postoperative auditory and language development [7-10]. Conversely, improvement after very late implantation in adolescents or young adults is generally expected to be slower, less predictable, and more dependent on rehabilitation intensity, prior communication mode, and duration of auditory deprivation [9,10].

The vascular finding in the eldest sister also has practical surgical relevance. A high-riding dehiscent jugular bulb with intratympanic protrusion influenced the choice of the implanted side. Careful review of preoperative temporal bone computed tomography remains essential in cochlear implantation, particularly to identify vascular variants that may increase surgical risk or modify the operative strategy [11,12].

This study has several limitations. It is a small retrospective family series with short follow-up. Genetic confirmation was not obtained. Speech therapy follow-up, standardized postoperative audiological testing, and validated language outcomes were incompletely documented. The postoperative CAP categories were estimated rather than prospectively collected. Larger multicenter reports with longer follow-up and standardized outcome measures are needed to better define cochlear implantation outcomes in patients with Chudley-McCullough syndrome.

CONCLUSION

In familial profound sensorineural hearing loss, the association of corpus callosum abnormalities, parasagittal frontal polymicrogyria, heterotopia, and arachnoid cysts on brain magnetic resonance imaging should raise suspicion for Chudley-McCullough syndrome, even before molecular confirmation. When the cochlear nerves are visible and inner-ear anatomy is favorable, cochlear implantation may be considered on an individualized basis. Genetic confirmation remains desirable, but auditory rehabilitation should not necessarily be delayed solely because molecular testing is unavailable. Functional conclusions remain preliminary in this series because follow-up was short and outcome assessment was retrospective.

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Declarations
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Conflicts of interest: The authors declare no conflicts of interest.

Ethics and consent: Written informed consent for publication of anonymized clinical and imaging data was obtained from the adult patient and from the legal guardians of the minor patients.

Data availability: The data supporting this case series are available from the corresponding author upon reasonable request, within the limits of patient confidentiality.

REFERENCES

1. Korver AMH, Smith RJH, Van Camp G, Schleiss MR, Bitner-Glindzicz MAK, Lustig LR, *et al.,* Congenital hearing loss. Nat Rev Dis Primers. 2017; 3:16094. doi:10.1038/nrdp.2016.94.
2. Chudley AE, McCullough C, McCullough DW. A syndrome of sensorineural deafness and structural brain abnormalities. Clin Genet. 1997; 52:287-293.
3. Doherty D, Chudley AE, Coghlan G, Ishak GE, Innes AM, Lemire EG, *et al.,* GPSM2 mutations cause the brain malformations and hearing loss in Chudley-McCullough syndrome. Am J Hum Genet. 2012;90(6):1088-1093. doi: 10.1016/j.ajhg.2012.04.008.
4. Ozdemir M, Dilli A. Chudley-McCullough syndrome: imaging findings and review. J Clin Imaging Sci. 2018; 8:45. doi: 10.4103/jcis.JCIS_39_18.
5. Alter NE, Introcaso K, Nunez C, *et al.,* Pediatric cochlear implants in Chudley-McCullough syndrome: report of two cases. Cureus. 2024;16(3): e55440. doi:10.7759/cureus.55440.
6. Forli F, Fiacchini G, Berrettini S, *et al.,* Cochlear implant in a subject affected by the Chudley-McCullough syndrome. Cochlear Implants Int. 2019;20(4):217-221. doi:10.1080/14670100.2019.1595875.
7. Archbold S, Lutman ME, Marshall DH. Categories of auditory performance. Ann Otol Rhinol Laryngol Suppl. 1995; 166:312-314.
8. Archbold S, Nikolopoulos TP, O'Donoghue GM, Lutman ME. The inter-user reliability of categories of auditory performance. Br J Audiol. 1998;32(1):7-12. doi:10.3109/03005369809076561.
9. Sharma A, Dorman MF. Central auditory development in children with cochlear implants: clinical implications. Adv Otorhinolaryngol. 2006; 64:66-88.
10. Kral A, Sharma A. Developmental neuroplasticity after cochlear implantation. Trends Neurosci.

2012;35(2):111-122. doi: 10.1016/j.tins.2011.09.004.

11. Tudose RC, Rusu MC, *et al.,* Anatomical variations of the jugular bulb: a comprehensive review. Medicina (Kaunas). 2024;60(9):1408. doi:10.3390/medicina60091408.
12. Sarioglu FC, Sarioglu O, Aydin E, *et al.,* Variations of the vascular canals in cochlear implant candidates. Int J Pediatr Otorhinolaryngol. 2019; 123:123-127. doi: 10.1016/j.ijporl.2019.05.001.