

# Chudley McCullough syndrome

The Chudley-McCullough Syndrome (CMS) is a rare autosomal-recessively inherited disorder caused by mutations in the GPSM2 gene, characterised by deafness and brain anomalies. The purpose of this paper is to report about a case of cochlear implant (CI) procedure in a subject affected by CMS.

**METHODS:** A 31-year-old subject affected by CMS referred to our centre requiring an evaluation for a CI, as the results with her hearing aids, which she had been using since she was 2-years-old, were unsatisfactory. A profound bilateral sensorineural hearing loss was pointed out. Pure tone audiometry in free field with hearing aids and speech perception results were poor. The subject was counselled about the surgical procedure and the surgery was performed with no complications.

**RESULTS:** The cochlear implant was switched on 22 days after surgery and the subject began speech therapy training. After 1 year, hearing and speech perception results were satisfactory. The hearing threshold in free field with the CI was around 30 dB, and the open set speech perception score reached 55% in silence.

**CONCLUSIONS:** The reported case demonstrates that CI is a feasible and safe procedure in subjects with CMS. Furthermore, since satisfactory hearing and speech perception results were achieved we recognise that cochlear implant should be considered the best option for hearing restoration in subjects with CMS and profound sensorineural hearing loss <sup>1)</sup>.

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<sup>1)</sup>

Forli F, Fiacchini G, Bruschini L, Caniglia M, Berrettini S. Cochlear implant in a subject affected by the Chudley-McCullough Syndrome. Cochlear Implants Int. 2019 Mar 25:1-5. doi: 10.1080/14670100.2019.1595875. [Epub ahead of print] PubMed PMID: 30907716.

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