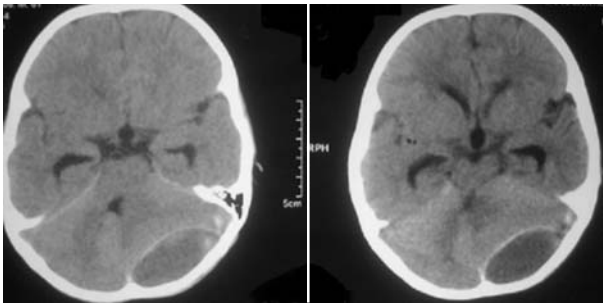
Late Decompensation after a Prolonged Lucid Interval in Chronic Posterior Fossa Extradural Hematoma

A 6-year-old boy presented in an unconscious state with history of holocranial headache and vomiting of two days duration. There was no history of seizures or fever. He was afebrile but tachypneic, was localizing with both upper limbs, with no eye opening or verbal response (Glasgow Coma Scale of E1M5V1). Pupils were equal in size and reacting to light. Both plantars had withdrawal response. There was a history of fall, 3 weeks ago, following which he lost consciousness for 20 minutes and was then apparently normal except for complaints of occipital pain and was treated conservatively. Computed tomography (CT) scan revealed a left-sided hypodense extra-axial mass in the posterior fossa compressing the left cerebellar hemisphere and brain stem effacing the perimesencephalic cistern with 4th ventricular shift (***Fig* 1**). His hematological parameters were normal. He underwent left cerebellar burr hole and evacuation of the hematoma under general anesthesia. Intraoperatively, altered liquid blood under pressure was drained and underlying dura was normal. A diagnosis of chronic extradural hematoma (EDH) was made. He regained consciousness the following day and was neurologically normal at follow-up, two months later.

One of the well-described classical presentations of patients with acute extra dural hematoma is a history of transient loss of consciousness following injury with subsequent recovery for a variable period before lapsing back into unconsciousness. This period of transient neurological recovery is called the lucid interval and occurs in 14-21% of patients with extra dural hematoma [1]. While there is no consensus on how long this period may span, it has been described by Ganz as lasting from a few hours to a few days [2]. The length of the lucid interval will be longer if the accumulation of blood is slow, as in venous origin of bleed or if there is significant shunting of blood outwards through the epidural veins [2].

Given the absence of fresh bleeding (in imaging) in this case the probable pathophysiology is expansion of the initial EDH by fluid, flowing in down an osmotic gradient, like in a chronic subdural hematoma leading to brain stem compression and 4th ventricular shift.



(*a*) (*b*)

FIG. 1 *Axial CT scan images (a,b) showing a hypodense biconvex extra-axial collection in the posterior fossa on the left side with 4th ventricular shift, cisternal effacement and rounding of the third ventricle.*

Posterior fossa constitute around 4-13% of all extradural hematomas [3], and sudden worsening after an initial hypo-symptomatic period has been reported [4]. This worsening has been reported only in the acute stage, though 11% of all extradural hematoma become chronic over time [1]. To predict patients likely to require surgery, Bozbuga, *et al*., [5] noted that acute posterior fossa EDHs having perimesencephalic cisternal effacement and 4th ventricular shift were more likely to require intervention. This patient too demonstrated both these features on imaging, though the hematoma had become chronic.

This case was unusual because there was delayed decompensation in the chronic stage after a prolonged lucid interval, and that the expansion and mass effect was not related to progressive bleed. The importance of continued close observation and follow-up (particularly in children who cannot describe subjective symptoms accurately) in conservatively treated extra dural hematoma is emphasized as the symptom progression may be 'silent and slow' [4,5], with sudden deterioration.

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New Mutation Haplotypes in Non-Syndromic Hearing Loss

We report on a male Caucasian child of two years of age, who attended Early Intervention Clinic with history of speech delay. There was family history of hearing loss in paternal uncles and aunts. Physical examination was normal with appropriate development and non-dysmorphic features. Assessment of internal, middle and external ears revealed no abnormality. Pure tone audiometry revealed bilateral profound sensorineural hearing loss of more than 90db of both middle and high frequency sounds. Genetic study was performed by PCR sequencing of the DNA carrying coding exon 2 of the gene *GJB2* (*Connexin 26*) and its flanking sequences (235nt). It identified c.35delG and c.427delC mutations in the aforesaid gene. Analysis of electrophoretogram revealed these two mutations in *Cis* position which has never been previously described in literature. He was subsequently fitted with hearing aids and also underwent hearing rehabilitation (speech therapy). At three years of age, he showed good improvement in his speech and had started using two to three word sentences.

Almost 80% of hereditary hearing loss is of nonsyndromic type [1]. Autosomal recessive, non-syndromic hearing loss and deafness (DFNB1) is characterised by non-progressive, mild to profound congenital hearing impairment that is detected at an early age. Though genetic linkage has demonstrated more than twenty hitherto unidentified genes in its causation, allelic mutation at *GJB2* locus has been reported in more than 90 percent of deafness cases [1,2]. Single nucleotide deletion of a Guanine residue from string of 6 guanine nucleotides from position 30 to 35 (c.del30G or c.del35G) is the most common identified mutation in Caucasian patients. Clinical heterogeneity of GJB2 mutations support the role of other unidentified genetic factors and environmental effects [3].

Assignments of alleles to the chromosomes (Haplotype) yields important information regarding recombination. Recombination events are marked in both family- and population-based linkage analysis to pinpoint disease causing mutations. Often haplotypes (several *cis* acting sites) are required to produce a phenotype, affecting gene products. Haplotypes by affecting gene product can either increase or decrease the severity of the phenotype [4]. Though identification of c.427delC as a *Cis* position haplotype could not be attributed to have any effect on phenotypic expression in our case as c.35delG alone has the potential to cause mild to profound hearing loss, similar reporting of further case or series with phenotypic attributes of this *cis* position haplotypes would help formulating the particular role of this haplotype (c.del35G and c.427delC) in modifying clinical expression of congenital non-syndromic hearing loss. We hope this case highlights the importance of genetic testing in children with sensorineural deafness as it helped us to pick up this novel haplotype which has not been previously reported in medical literature.

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