
ISSN: 3068-8965
Journal of Pediatric Advance Research

ATHENAEUM SCIENTIFIC PUBLISHERS

Case Report

Brain Abnormalities in "Pure" Trisomy 20: A Case Report

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Citation: Sintini M, et al. Brain Abnormalities in "Pure" Trisomy 20: A Case Report. J Pediatric Adv Res. 2026;5(1):1-6. https://doi.org/10.46889/JPAR.2026.5103

Received Date: 24-12-2025

Accepted Date: 19-01-2026

Published Date: 25-01-2026

Abstract

Background: Trisomy 20p, also known as chromosome 20 trisomy, is a rare genetic disorder involving developmental delays, mild intellectual disability, motor skill impairments, hypotonia and speech difficulties.

Methods and Findings: The child with pure trisomy 20 reported here presented with ideomotor dyspraxia, spatial discrimination deficits and impaired fine motor skills. Neuroimaging revealed diffuse developmental brain hypoplasia with specific underdevelopment of the mesencephalic tegmentum, resulting in the characteristic "hummingbird or penguin sign." This sign was commonly observed in degenerative diseases and, recently, in idiopathic normal pressure hydrocephalus of adults. For many reasons, however, this child's findings cannot be attributed to idiopathic normal pressure hydrocephalus. Furthermore, the child had a history of focal seizures requiring antiepileptic drug therapy.

Conclusion: This child may be the first reported case of trisomy 20 presenting with diffuse developmental brain hypoplasia and the characteristic "hummingbird sign" on neuroimaging, as well as focal epileptic seizures.

Keywords: Trisomy 20; Development; Brain Hypoplasia; Hummingbird Sign; Epilepsy

Introduction

Trisomy 20p, also known as chromosome 20 trisomy, is a rare genetic disorder resulting from the duplication of all or part of the short arm of chromosome 20. Many reported patients have partial trisomy 20, while only a few have complete de novo trisomy. The severity of the clinical phenotype depends on the size of the chromosome 20p duplication; larger duplications result in a more disabling disorder. The most representative clinical phenotype includes specific facial features, developmental delay with mild cognitive impairment, hypotonia, poor coordination of gross and fine motor skills and speech impairment [1-5]. Spinal, cardiac and renal abnormalities may also be present. This condition should be distinguished from mosaic trisomy 20, which is more common and involves three copies of chromosome 20 in some cells. While some developmental defects are shared with pure trisomy, mosaic trisomy more often presents with learning disabilities and behavioral disturbances such as isolation, social difficulties, temper outbursts, panic attacks, irritability, depressed mood and self-injury and suicidal ideation [6,7]. Structural brain abnormalities have rarely been demonstrated in individuals with trisomy 20p. These abnormalities include a posterior fossa anomaly that mimics Dandy-Walker malformation [8]; a left temporal arachnoid cyst and a small pineal gland cyst [4]; and congenital bilateral perisylvian polymicrogyria, as well as corpus callosum (CC) and cerebellar hypoplasia, in an adult male with trisomy 20p/monosmy 18p [9]. There can also be no detectable abnormality [5]. Brain structural defects are also rare in mosaic trisomy 20, although there have been more reported cases. Noteworthy, an arachnoid cyst in the basal cistern that mimicked a Dandy-Walker Malformation (DWM) and micrencephaly with associated neuronal subependymal heterotopies have been documented [10,11]. Curiously, epileptic aphagia with abnormal Electroencephalographic (EEG) recordings and normal development but delayed speech was reported alongside global atrophy on neuroimaging in a single case [12]. Here, we report

a male child with pure trisomy 20p who presented with epileptic seizures and various structural brain abnormalities, curiously including the appearance of a typical hummingbird sign on neuroimaging.

Case Report

A six-year-old child was referred for a neurological evaluation due to diffuse developmental delay. He exhibited no specific phenotypic abnormalities, except for defective physical growth. Neurologically, he was hypotonic and exhibited motor clumsiness, a clear learning disability and delayed formal, conceptual and symbolic language development. The rest of the neurological examination was normal. Interestingly, he had been suffering from focal seizures since age four, that were satisfactorily controlled with oral carbamazepine. Due to his mental and neurological condition, as well as his early-onset focal epilepsy, genetic testing was performed using array Comparative Genomic Hybridization (aCGH). This testing revealed complete trisomy of the short arm of chromosome 20, indicating a diagnosis of trisomy 20. Since this case is from an old archive, the complete genetic testing results are unavailable. However, the child exhibited all the phenotypic characteristics of this aneuploidy, including a ventricular cardiac septal defect and upper thoracic vertebral dysmorphism. Both cranial Computed Tomography (CT) and Magnetic Resonance Imaging (MRI) of the brain revealed various abnormalities, some of which not previously described. The cortical convolutions were normal, but the frontal, temporal and parietal gyri appeared moderately hypoplastic, with deeper sulci and an increased volume of the arachnoid space. This was particularly evident at the temporal lobe poles, resulting in cystic-like appearance. The insular arachnoid spaces were abnormally dilated on both sides due to defective growth of the fronto-temporo-parietal parenchymal tissue. The hemispheric white matter had a uniformly reduced volume, yet it showed normal signal. The corpus callosum was extremely thin and arch-shaped. The ventricular system was abnormally dilated, particularly the third ventricle, with consequent partial hypoplasia of the thalami. The head of the caudate nucleus and the putamen appeared to be less involved. The fourth ventricle exhibited normal morphology, yet was dilated, likely due to dorsal brainstem hypoplasia and small megacisterna magna. Available images did not consent to adequately evaluated the permeability of the aqueduct of Sylvius, which seemed mildly dilated. The bulbo-medullary junction appeared normal. The sella turcica was particularly tight and deep and contained a very small hypophysis. There was no heterotopic neuronal mass (Fig. 1,2).

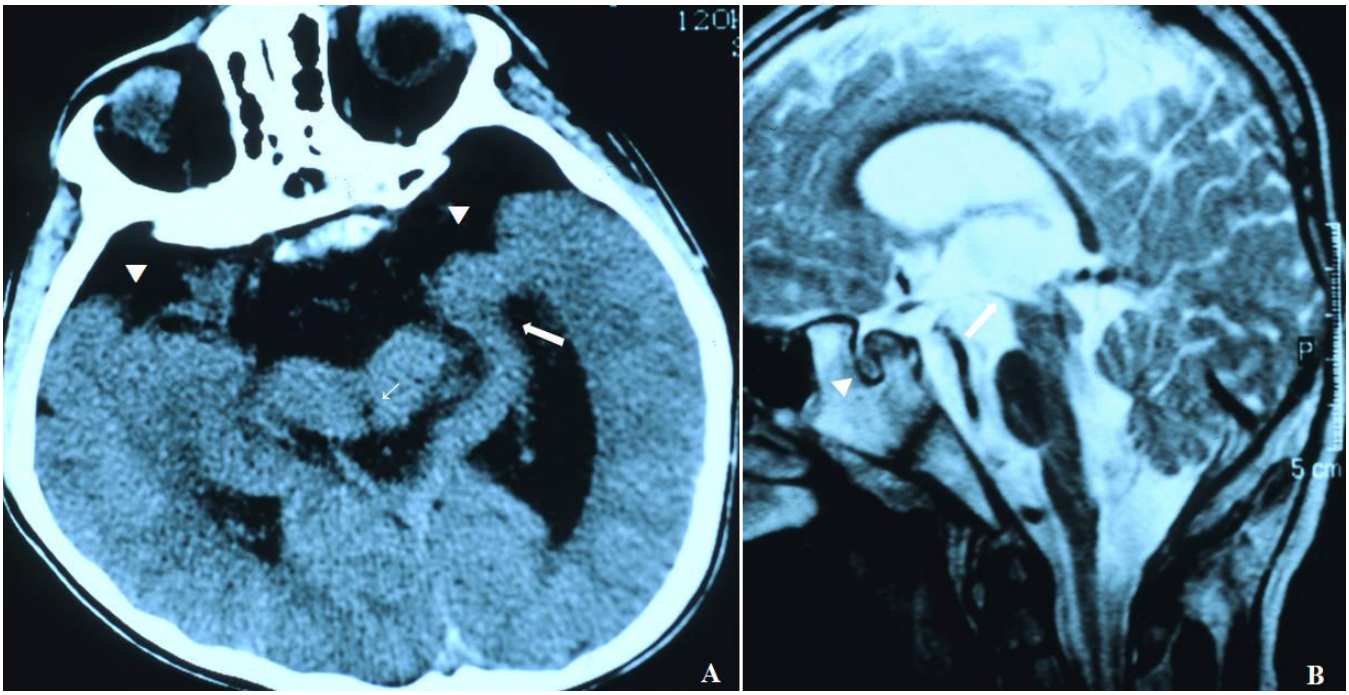

Figure 1: The brain CT scan (Panel A) shows that the dilatation of the temporal pole's arachnoid spece is not a true cyst (filled white stars). The mesencephalon is underdeveloped, especially the tegmentum. The acqueduct of Sylvius is slightly dilated (thin white arrow). The temporal horn is widely dilated and the hippocampal formation is hypoplastic (large white arrow); Panel B (sagittal MRI T2 SE). The hummingbird sign is clearly evident (large white arrow), as is the small sella turcica with an almost absent hypophysis (white filled star). Note the thin arciform CC, ponto-bulbar hypoplasia, dilated basal cisterns, dilated fourth ventricle and slightly upwardly dislocated cerebellar vermis by a small megacisterna magna.

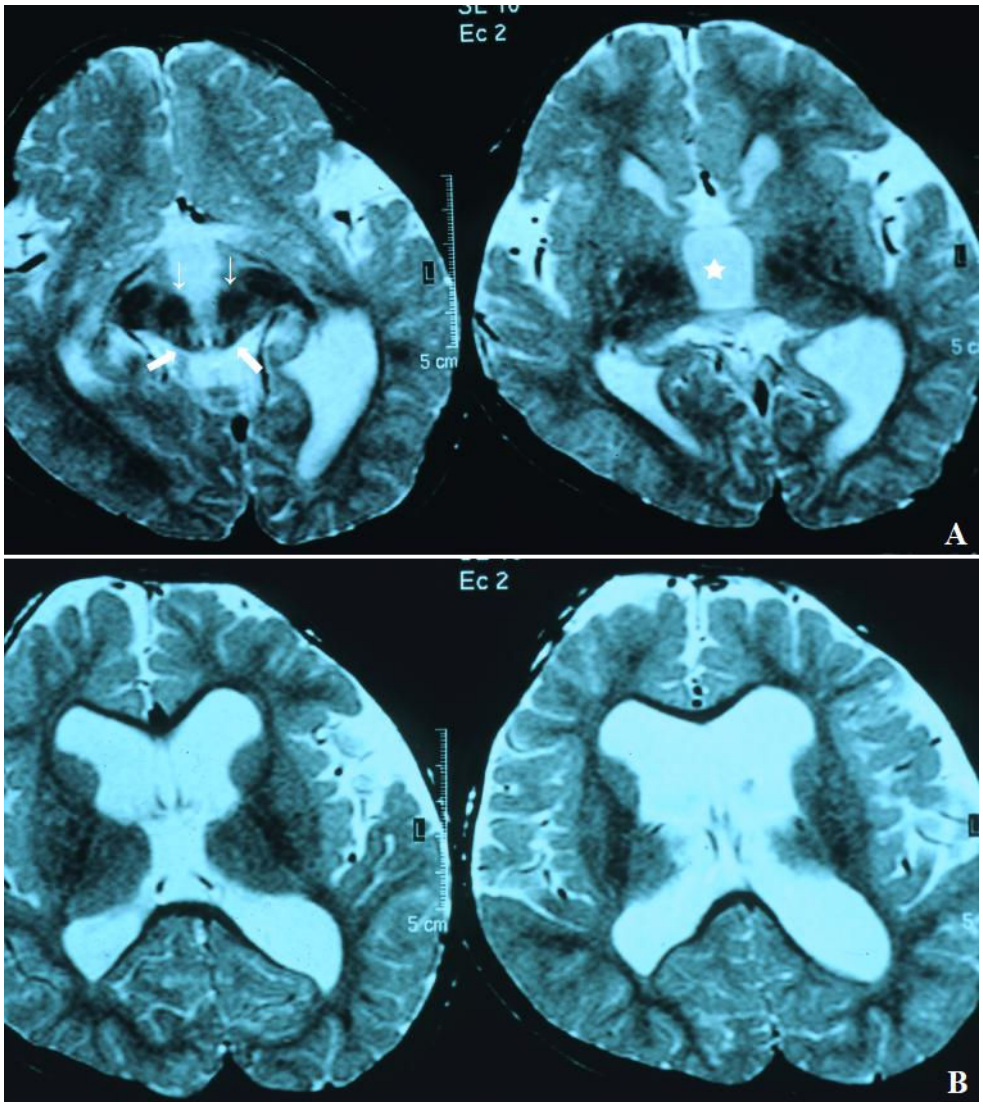

Figure 2: Axial MRI T2 SE (Panel A and B). The entire supratentorial ventricular system is dilated, particularly the third ventricle (filled white star). The hypoplasia of the mesencephalic tegmentum is evident (large white arrows) and the ventral mesencephalon and red nuclei are well preserved and recognizable (thin white arrows). Fronto-temporal hypoplasia with enlarged subarachnoid spaces is evident, exposing the insular cortex. The CC does not form an acute angle, as can be seen in Panel B. The thalami are particularly underdeveloped.

Discussion

Overall, few studies have examined brain neuroimaging in trisomy 20 aneuploidy, including pure and mosaic forms. Various abnormalities have been demonstrated, including temporal or pineal gland cysts, perisylvian polymicrogyria, posterior fossa anomalies resembling DWM, microencephaly with subependymal heterotopias and global atrophy [4,5,8-13]. The neuroimaging study of the child reported here revealed various previously undescribed structural brain abnormalities in pure trisomy 20 that raise interesting points of discussion.

In this reported child, the dilatation of the entire supratentorial ventricular system, particularly the third ventricle, as well as the arachnoid spaces, especially those of the temporal poles and insular regions and all the basal cisterns, along with the thin, arch-shaped CC, may suggest a severe, hypertensive defect in the circulation of Cerebrospinal Fluid (CSF). However, there is no apparent obstruction to its circulation. Though not clearly demonstrated by few available neuroimages, the aqueduct of Sylvius is likely open due to a dilated fourth ventricle. Additionally, the extreme dilation of the temporal and insular arachnoid spaces on both sides, as well as the significant enlargement of the third ventricle, are difficult to explain by hydrocephalic hypertension, especially given the absence of CSF obstructive signs. In the posterior fossa, hypoplasia of the dorsal pontobulbar brainstem and a likely megacisterna magna may contribute to enlargement of the fourth ventricle. These are not primary signs of hypertensive

hydrocephalus and the same is true for dilated basal cisterns. The extreme thinning of CC for the patient's age may due to Cerebrospinal Fluid (CSF) hypertension, but it is most likely a true developmental malformation. Thin and arciform CC is a common anomaly in several genetic disorders. Regarding this reported patient, callosal hypoplasia has only been observed in conjunction with other complex brain malformations in a patient with trisomy 20p/monosomy 18p [9]. Therefore, we suggest that it may be a new malformative aspect of pure trisomy 20p. Additionally, the lateral ventricle walls had a squared appearance, similar to that seen in dysgenetic conditions. There was also no evident transependymal CSF reabsorption. Therefore, it is difficult to conclude that the patient had hydrocephalus due to CSF hypertension.

Due to the morphology of the median thalamus, the disproportionate enlargement of the third ventricle itself cannot be a malformative cyst. It may be due to a genetic anomaly. Similarly, dilatation of the arachnoid space at the temporal poles cannot be considered a malformative cyst, as Bartolini suggested in a fetus with partial trisomy [4]. Overall, these anomalies demonstrate moderate to severe diffuse brain hypoplasia and abnormal development of the intracerebral and extracerebral CSF spaces.

Notably, the small, pointed beak of the ventral mesencephalic tegmentum-the so-called "hummingbird or penguin sign" may have intriguing implications. This sign is commonly associated with Progressive Supranuclear Palsy (PSP), Parkinson's disease (both typical and atypical forms) and multiple system atrophy in adults. There, it is often associated with midbrain atrophy, dilated cisterns and thinning of the quadrigeminal plate, as well as third ventricular dilatation. However, it has also been observed in idiopathic Normal Pressure Hydrocephalus (iNPH) in adults [14,15]. The cause has not yet been conclusively explained. The MRI diagnosis of iNPH in adults includes the following: dilatation of the temporal horns; acute callosal angle; abnormal signal of the periventricular white matter; asymmetric enlargement of the sulci; enlargement of the subarachnoid spaces with dilation of the Sylvian fissure; callosal thinning; and a wider third ventricle. A variable number of affected patients show the hummingbird sign. Vascular compression associated with third ventricle dilatation has been proposed as an explanation for morphological changes in the mesencephalon [15]. However, an alternative theory claims that a tortuous posterior circulation causes displacement of the floor of the third ventricle and concave transformation of the upper mesencephalon [16]. The hummingbird sign in degenerative diseases such as typical and atypical Parkinson's disease, PSP and multiple system atrophy is instead attributed to ongoing local atrophy. Some neuroimaging features characteristic of iNPH in adults were present in this child with trisomy 20. However, the acute callosal angle, a cardinal feature of iNPH, was not present. This may depend on the retrospective nature of our case study and the small number of available neuroimages; however, the CC did not appear to form an acute angle. Conversely, the third ventricle was unusually large and may have contributed to anterior quadrigeminal displacement, along with dilated anterior cisterns. In our case, the absence of abnormal signals in the periventricular white matter and the lack of a recognizable acute callosal angle make diagnosing iNPH according to adult criteria difficult. Furthermore, the defining neuroimaging features of iNPH in rare cases of affected children is ventricular dilation accompanied by disproportionate narrowing of the subarachnoid space and cortical sulci. Therefore, this child cannot be described as having typical infantile iNPH, similarly to adults. The finding of the hummingbird sign alongside a dilated third ventricle, similar to that seen in adult iNPH, remains difficult to explain. However, it is noteworthy that hypoplasia involves the mesencephalic tegmentum, the dorsal pontobulbar brainstem, the sella turcica, the hypophysis and the entire supratentorial brain, especially the white matter, as well as a thin corpus callosum. These characteristics demonstrate generalized hypoplastic development of the entire brain and its connecting fibers, similar to what occurs in many dysgenetic conditions. These abnormalities may contribute to neuroimaging dysmorphisms similar to iNPH. Therefore, we suggest a malformative condition involving multiple anomalies related to trisomy 20 aneuploidy. This child may be the first pediatric case of hummingbird sign alongside mesencephalic quadrigeminal dysmorphism that is identical to the hummingbird sign observed in adult's degenerative disease and iNPH. This case describes new brain abnormalities in young individuals with pure trisomy 20. Interesting, studies of families with iNPH have shown how difficult it is to identify specific genetic substrates [17,18]. One interesting example is the CCNO gene defect in Primary Ciliary Dyskinesia (CILD29, OMIM #615872) in the Ceno mouse. This mutation results in abnormal and immotile cilia in ependymal multiciliated cells, leading to impaired CSF recirculation and subsequent iNPH [19].

Conclusion

Finally, the child reported here with pure trisomy 20 experienced true partial motor seizures with focal bioelectrical irritation. He was successfully treated with carbamazepine. He may be the first patient with trisomy 20p to have focal epilepsy. Except for a 29-year-old patient with trisomy 20p/monosomy 18p who had focal tonic seizures and a child with mosaic trisomy who

exhibited questionable epileptic aphagia with background slowing on EEGs and was on antiepileptic drug therapy, epilepsy has never been observed in patients with pure trisomy 20.

Conflict of Interest
The authors declared no potential conflicts of interest with respect to the research, authorship and/or publication of this article.

Funding Statement
This research did not receive any specific grant from funding agencies in the public, commercial or non-profit sectors.

Data Availability Statement
Data and Materials are available from the authors upon reasonable request.

Ethical Statement
This study was conducted in accordance with the ethical standards of the Helsinki Declaration of the World Medical Association. Furthermore, the manuscript did not need the approval of the Ethical Committee of our University Administration as this is not a requirement for the publication of a single case provided that it is of definite interest to the scientific community (Regulations of the Ethical Committee of "Area Vasta Emilia Nord", Italy, approved on September 22, 2020).

Informed Consent Statement
The informed consent has been obtained verbally and the patient anonymity preserved.

Authors' Contributions
Conceptualization: E.D.G.; M.S.; Data collection and analysis: E.D.G.; M.S.; Data curation: M.S.; Investigation: E.D.G.; Writing-original draft: E.D.G.; M.S.; L.R.B.; Writing-review and editing: All authors.

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