Perspective
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Short Note on Hajdu-Cheney Syndrome

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Perspective

Hajdu-Cheney Syndrome (HCS) is an extremely rare genetic disorder. It is registered in the OMIM project database under the reference number 102500 and in ORPHANET under the reference ORPHA955. This disease, which belongs to the osteolysis syndromes group, primarily affects connective tissue. Acro-dento-osteo-dysplasia, acroosteolysis with osteoporosis and changes in the skull and mandible, arthro-dento-osteo-dysplasia, and serpentine fibula–polycystic kidney syndrome are other names for it. It is caused by a heterozygotic mutation of the NOTCH2 gene on chromosome 1p13-p11 and is inherited autosomally dominantly, though descriptions of cases with sporadic mutations can be found. This disease affects less than one person in a million (1/1,000,000).

N. Hajdu first described the disease in 1948, and D. Cheney completed the description in 1965. Since then, approximately 100 cases have been reported in the scientific literature, allowing for the identification of a number of common features shared by all patients, such as phenotypic variability, age-dependent progression, and the presence of generalised osteoporosis and acroosteolysis of distal phalanges, among other clinical manifestations.

Because of the variability in NOTCH2 expression, phenotypic variability exists; as a result, patients with this disease may present with clinical differences. Furthermore, because this disease has such a broad and specific clinical spectrum, it may be difficult to encounter it all in a single patient. Because this disease is degenerative, the clinical manifestations worsen over time, with the onset of many changes ranging from early childhood to late adulthood. The osteolysis of distal phalanges and generalised osteoporosis found in all HCS cases are accompanied by a series of clinical manifestations that, as previously stated, vary between patients.

These differences include cranial alterations such as dolichocephaly, delayed suture closure, the presence of multiple wormian bones, the absence of frontal sinuses, a thickened dome of the skull, occipital prominence, bathrocephaly, elongated sella turcica, and micrognathia, which may lead to complications such as basilar invagination, hydrocephalus, and syringomyelia; facial alterations such as hypertel Other clinical manifestations may include delayed motor development, hearing loss, voice changes, congenital heart disease, respiratory, renal, and digestive system changes, plantar ulcers, and hernias.

Although the initial diagnosis is based on the observation of external appearance and radiological findings, genetic sequencing is used to reach a definitive diagnosis. Certain overlapping features with other diseases, such as scleroderma, sarcoidosis, progeria, pycnodysostosis, Whyte-Hemingway, Winchester, and Alagille syndrome, may necessitate inclusion in the differential diagnosis on occasion. There is currently no definitive or effective pharmacological treatment for HCS, though research is being conducted in this area. At the moment, treatment for this disease is focused on managing complications and underlying issues in order to improve the patient's quality of life and life expectancy. There are currently over 7000 rare diseases in the world, with only 800 having limited scientific understanding. Rare diseases, in general, and Hajdu–Cheney syndrome in particular, have a low prevalence and few documented cases. The population sample for the study is dispersed, with a variable phenotype, little-described clinical symptoms, and a different evolution [1-5].

References

1. Pittaway, James FH, Christopher Harrison, Yumie Rhee snd Muriel Holder-Espinasse, et al. "Bisphosphonate therapy for spinal osteoporosis in Hajdu-Cheney syndrome–new data and literature review." *Orphanet J Rare Dis* 13 (2018): 1-7.
2. Cortés-Martín, Jonathan, Juan Carlos Sánchez-García, Beatriz Piqueras-Sola and Raquel Rodríguez-Blanque, et al. "Hajdu-Cheney Syndrome: Report of a Case in Spain." *Diagnostics* 12 (2022): 566.
3. Vollersen, Nele, Irm Hermans-Borgmeyer, Kerstin Cornils and Boris Fehse, et al. "High Bone Turnover in Mice Carrying a Pathogenic Notch2 Mutation Causing Hajdu-Cheney Syndrome." *J Bone Miner Res* 33 (2018): 70-83.
4. Falls, Cody J, Paul S. Page and James A. Stadler. "Craniospinal Surgery in Hajdu-Cheney Syndrome: A Review of Case Reports." *Cureus* 13 (2021).
5. Graversen, Lise, Mette Moller Handrup, Melita Irving and Hanne Hove, et al. "Phenotypic presentations of Hajdu-Cheney syndrome according to age–5 distinct clinical presentations." European J Med Genet 63 (2020): 103650.

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