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Craniofacial fibrous dysplasia: case report of uncommon genetic defect in young adult

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ABSTRACT

Fibrous dysplasia is uncommon genetic defect in which normal bone and marrow are replaced with fibro-osseous tissue, leading to bone deformities, pain fractures and functional impairments. The case presents 27 years old women that was diagnosed with sepsis in the course of catheter-related infection and treated at nephrology department. She was diagnosed with chronic kidney disease that started during pregnancy and originated due to steroid-resistant nephrotic syndrome. During the hospitalization, a CT scan of the head was performed because of facial deformation. Based on CT scans of head, craniofacial fibrous dysplasia was confirmed.

Materials and Evidence: A literature review was conducted using the PubMed database.

Keywords: craniofacial fibrous dysplasia, fibrous dysplasia, rare bone disease, bone neoplasms, McCune-Albright syndrome

INTRODUCTION

Fibrous dysplasia is uncommon disease that can appear spontaneously through genetic mutations which are nonheritable. The complexity of this disease lies in replacement of bone and marrow with fibrous tissue. Due to replacement of normal bone with fibro-osseous lesions filled with abnormally proliferating osteoprogenitor cells. Pathophysiology of this replacement comes from activation in GNAS gene, which encodes alfa-subunit of the stimulatory *Gs*G-coupled protein receptor. In result receptor gets activated constantly and leads to production of cyclic-AMP, this Gas signaling causes impairing normal differentiation of bone marrow sromal cells and resulting in disorganized bone matrix formation. [1] [2]

From all bone tumors fibrous dysplasia accounts for 5% to 7%. Depending one number of bones affected we can distinguish: monostotic fibrous dysplasia or polyostotic fibrous dysplasia. From these two variants in craniofacial dysplasia monostotic is more common one, it affects 56% of patients, 47% of patients had polyostotic disease and small portions of all patients had McCune-Albright syndrome. [1][2]

Clinical manifestation of craniofacial fibrous dysplasia is highly variable and depends on localization of lesion's. Common symptoms include painless facial swelling, asymmetry, malocclusion, dental anomalies and occasionally cranial nerve compression. Some cases can present displacement of cranial skeleton, such as orbits that can present with visual disturbances, external or internal auditory canals deformations which can lead to hearing impairment, in some cases may occur congestion of nasal passages from pathologic deformation. In some cases there are no manifestations and patients remain asymptomatic through life, with incidental diagnosis, getting a computed tomography. [2][3]

Diagnosis of craniofacial fibrous dysplasia is primarily based on clinical features of patients. These features are complemented by comprehensive evaluation of skeletal, endocrine, dermatologic and soft tissue feature. Radiological image is gold standard for diagnostic due to craniofacial lesions presenting a characteristic "ground-glass" appearance, often with bone expansion and poorly demarcated margins. In monostotic cases, without systemic features, histological confirmation may be needed to differentiate craniofacial dysplasia from other fibro-osseous lesions. When clinical and radiographic findings are inconclusive, molecular testing for GNAS mutations in biopsy samples can assist in confirmation the diagnosis. [4]

CASE REPORT

The case presented is 27-year-old female, with chronic kidney disease at stage G5 (CKD) at secondary to nephrotic syndrome, currently undergoing maintenance hemodialysis. She presented herself to emergency department due to severe hypertension accompanied by headache, vomiting, weakness and dyspnea. After evaluation in the emergency department she was refered to nephrology for hemodialysis, where poor blood glow through the tunneled central venous catheter was noted (Qb 100ml/min). Due to the symptoms presented by the patient and catheter dysfunction, she was then referred to the nephrology department for catheter replacement.

She had been diagnosed with membranoproliferative glomerulonephritis in early childhood, at the age 6, it was confirmed by multiple renal biopsies (2004, 2010, 2013). Because of renal failure during her second pregnancy she started hemodialysis since 20th week of pregnancy, through a tunneled central venous catheter placed in the right internal jugular vein. After the pregnancy kidney failure persisted and she was chronically dialyzed.

During the first day (2 day after reporting to ER) of admission high fever (up to 39,4*C) with chills was noted, the remaining symptoms did not subside. Laboratory findings reveled signs of catheter-related bloodstream infection, including elevated CRP, procalcitonin, leukocytosis, hyperkalemia, elevated urea and creatinine. Chest X-ray showed cardiomegaly without pulmonary congestion and noted abnormal contours of the left seventh rib. Echocardiography later raised suspicion of mitral valve vegetation, suggesting possible infective endocarditis.

Date	**CRP (mg/L)**	**PCT (ng/mL)**
2025-01-07	85.49	22.789
2025-01-09	154.29	-
2025-01-11	70.97	19.766
2025-01-13	32.69	7.703
2025-01-20	5.23	1.234

Table 1. Blood test results of inflammatory parameters

During hospitalization, a diagnosis of catheter-related sepsis was made. The adherent tunneled catheter was removed using a high-pressure ballon and empirical antibiotic therapy was initiated, resulting in clinical improvement and decreased inflammatory markers. During hospitalization craniofacial deformities were observed, cranial CT was performed. Based on the CT scan suspicion for craniofacial fibrosus dysplasia was raised.

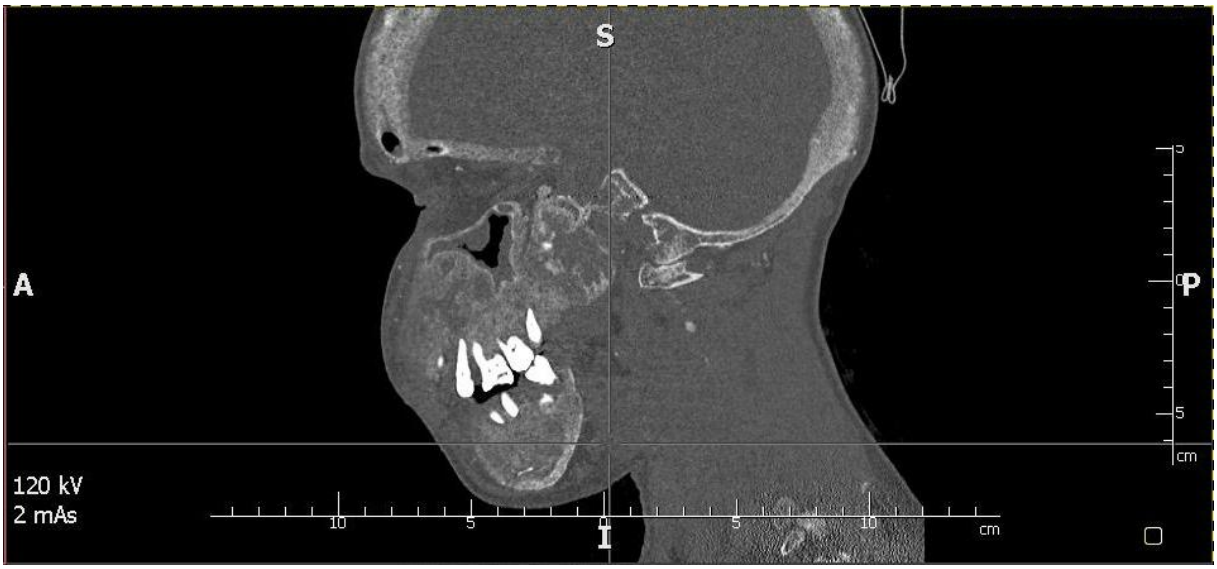

Figure 1. CT scan of head showing deformation of maxilla and mandibula.

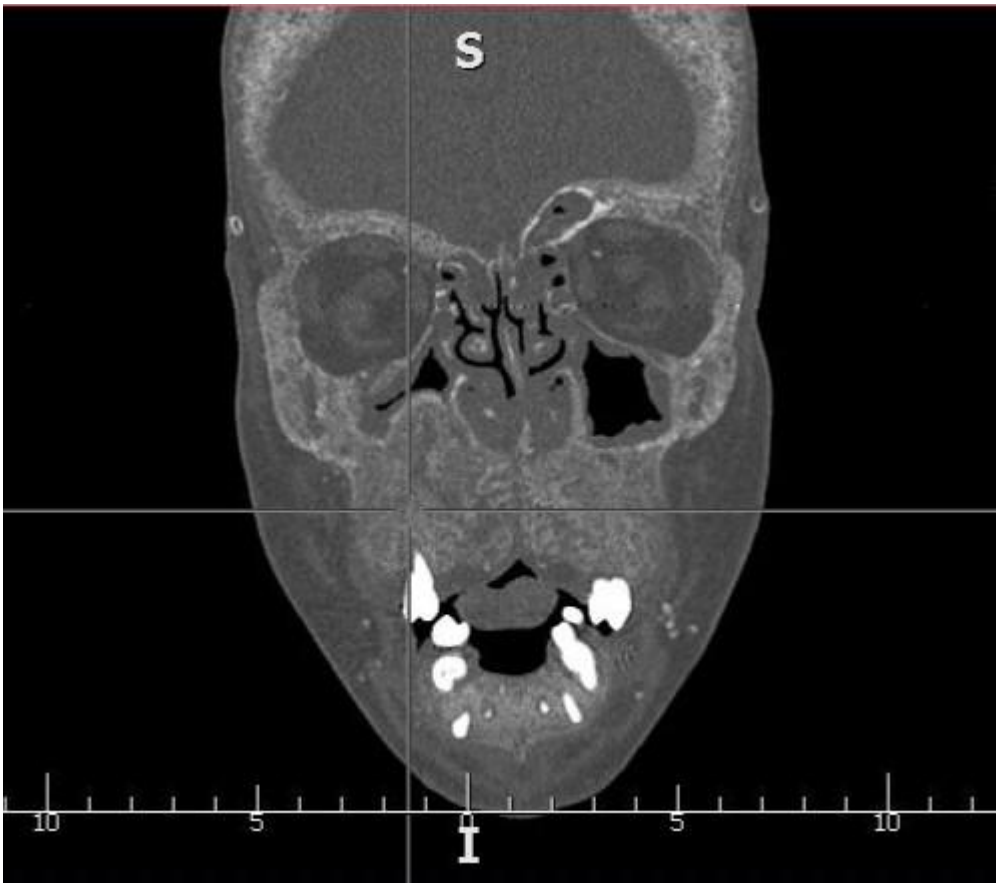

Figure 2. CT scan of head showing deformation of maxilla.

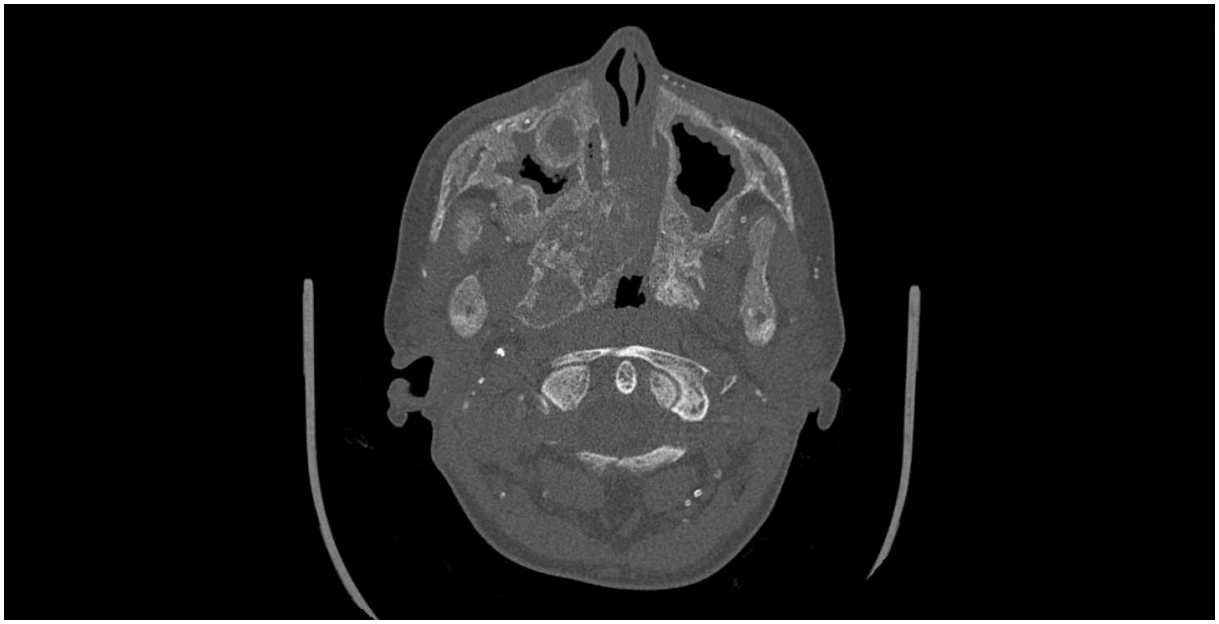

Figure 3. CT scan of head showing deformation of maxillary sinuses.

Figure 4. Catheter removed from the patient with visible bacterial biofilm.

Later during hospitalization transthoracic echocardiography raised suspicion of mitral valve vegetation, suggesting possible infective endocarditis. Patient declined and discharged herself against medical advice. She was referred to the genetic clinic; however, according to the physician overseeing her hemodialysis, she did not attend the appointment.

DISCUSSION

The presented case illustrates the incidental diagnosis of craniofacial fibrous dysplasia in a young adult that was undergoing evaluation for sepsis that was related to catheter. It shows that craniofacial lesions can often remain silent through life of patients, until deformity or complications arise. Consistent with literature, craniofacial involvement is common in fibrous dysplasia, present up to 90% of patients with polyostotic forms and may remain undetected until adulthood if asymptomatic [2, 3].

In case of this patients the asymmetry was observed, with no other symptoms and led to computer tomography [5]. The patient in this case presented clinical features and would qualify for genetic testing, best testing methods currently are PCR-based sequencing methods, detection rates are grater than 80% in lesional tissue. Detection of pathogenic *GNAS* mutation may be helpful but does not rule out diagnosis [7].

Menagement of craniofacial dysplasia remains challenging and individualized. Based on the international guidelines, treatment decisions depend on the lesion's size, symptoms and progression [4][6]. In asymptomatic patients or those with no progression of lesions, observations are preferred. On the other hand, if the patient presents symptoms such as vision disturbance, hearing impairment or cosmetic disfigurement are impacted, surgical intervention may be beneficial. However, the choice between conservative debulking and radical resection must balance recurrence risk with surgical morbidity [6]. In this case surgical management was not pursued due to patient's expressed preference to decline intervention.

Notably, our patient presented with multiple comorbidities, including end-stage renal disease, prolonged immunosuppression, and secondary endocrine disturbances, in this case hyperparathyroidism. In available literature there are some cases where fibrous dysplasia coexisted with hyperparathyroidism, it is not entirely sure it is coincidental or is it a rare family genetic syndrome [8, 9]. While severe infection and catheter-related endocarditis in not

directly causal for craniofacial fibrous dysplasia, it underscores the importance of holistic, multidisciplinary care in complex chronic disease settings.

CONCLUSION

In summary, this case highlights the diagnostic and therapeutic complexities of craniofacial fibrous dysplasia, the follow up after a visit to the clinical genetics center to which the patient was referred, could confirm whether the diagnosis using only imaging diagnostics was correct. The complexity of this patient emphasizes the need for a tailored multidisciplinary approach grounded in established diagnostic guidelines and evolving management strategies [6].

Disclosure:

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Author's contribution

Conceptualization: Konrad Haraziński, Weronika Goliat; methodology: Oliwia Sysło, Izabela Jastrzębska; software: Nikola Rubik; check: Maksym Gmur; formal analysis: Michał Gajewski; investigation: Barbara Sławińska; resources: Zuzanna Błecha; data curation: Izabela Jastrzębska; writing – rough preparation: Nikola Rubik, Maksym Gmur; writing – review and editing: Konrad Haraziński, Weronika Goliat, Oliwia Sysło; visualization: Michał Gajewski; supervision: Nicole Maryniak; project administration: Konrad Haraziński; receiving funding: Not applicable

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Institutional Review Board Statement

Not applicable.

Informed Consent Statement

Our work did not involve direct human subject research or obtaining their consent for participation in the study.

Data Availability Statement

As a review paper, our work does not present new data or analyses. Therefore, there are no specific databases or data availability to report. The information and findings presented in this review are based on previously published studies, which can be accessed through their resective sourses as cited in the reference section.

Conflicts of Interest Statement

The authors declare that there are no significant conflicts of interest associated with this research work.

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