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Case Study

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A CASE OF OP POISIONING DIAGNOSED AS ACHONDROPLASIA WITH SEVERE CALCIFIED AORTIC STENOSIS

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ABSTRACT

Background

Introduction: Achondroplasia is a genetic disorder characterized by abnormal bone growth, leading to disproportionate short stature. It is the most common form of dwarfism, with an incidence of approximately 1 in 15,000 to 40,000 live births.

KEYWORDS: Achondroplasia, Op Poisoning, Severe Calcific Aortic Stenosis.

Etiology and Genetics: Achondroplasia is caused by mutations in the FGFR3 gene, which encodes the fibroblast growth factor receptor 3. This gene mutation is autosomal dominant, meaning only one copy of the altered gene is sufficient to cause the disorder. Most cases (about 80%) result from de novo mutations, where there is no family history of the condition.

Pathophysiology

The FGFR3 mutation leads to an abnormality in the development and maintenance of bone and brain tissue. FGFR3 normally regulates bone growth by limiting the formation of bone from cartilage, especially in the long bones. The mutation causes this regulation to go away, resulting in the characteristic shortened limbs and other skeletal abnormalities.

Clinical Features

Individuals with achondroplasia have a distinctive appearance marked by.

- Disproportionately short stature, with an average adult height of around 4 feet.
- Short arms and legs, particularly the upper arms and thighs.
- Large head size with a prominent forehead (frontal bossing).
- Midface hypoplasia with a flattened nasal bridge.
- Spinal abnormalities, including lordosis and kyphosis.

Additional complications can include ear infections, sleep apnea, obesity, and spinal stenosis, which can cause pain and neurologic symptoms.

Diagnosis is primarily clinical, based on characteristic physical features. Genetic testing can confirm the presence of FGFR3 mutations. Prenatal diagnosis is possible through ultrasound and genetic testing if there is a known risk.

CASE PRESENTATION

A 52 year old male patient from meerut city, up presented to lokpriya hospital with a/h/o ingestion of op compound with vomiting, salivation, shortness of breath since 1 day; clinical finding suggestive of op poisoning and managed conservatively. routine blood investigations and workup was done and was within normal range.

ON PHYSICAL EXAMINATION

Patient was short statured with height-104 cm,arm span-145 cm, weight of the patient was 42 kg; ap diameter of chest- 12 cm, transverse diameter of chest- 18 cm; b/l chest expansion was reduced clinically diagnosis was made of achondroplasia Radiological imaging(chest x-ray) was done which showed cardiomegaly, both domes and on examination incidental murmur was detected in aortic area which was radiating to right carotid suggestive of valvular heart disease; to confirm it 2d echo was done which revealed severe calcific aortic stenosis.

CONCLUSION

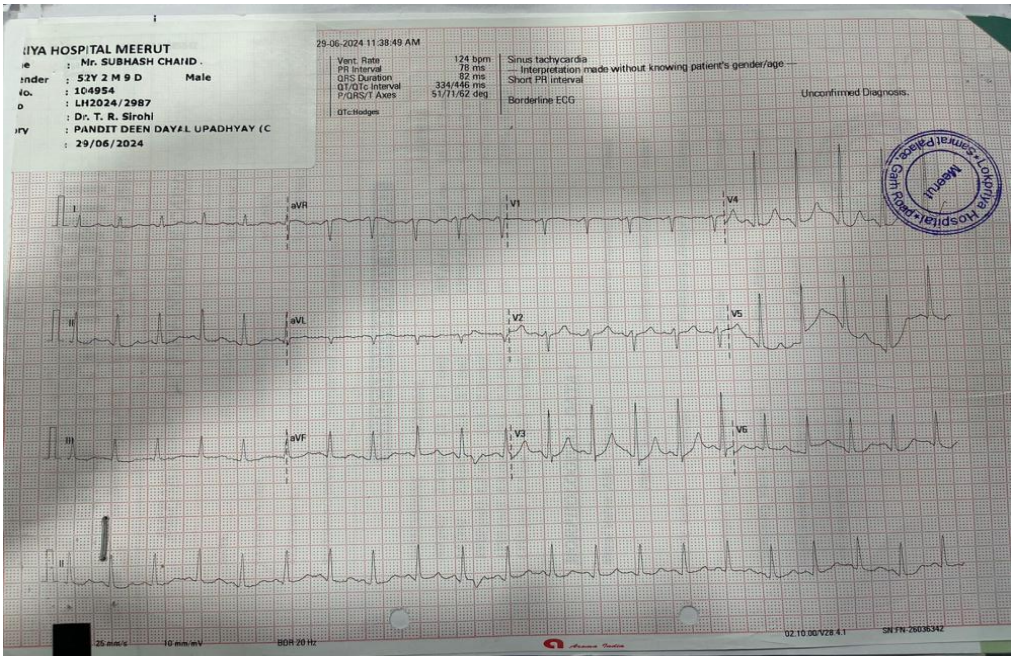
This case highlights the importance of early diagnosis and multidisciplinary care in managing achondroplasia. With appropriate medical support, individuals with achondroplasia can lead healthy, productive lives.

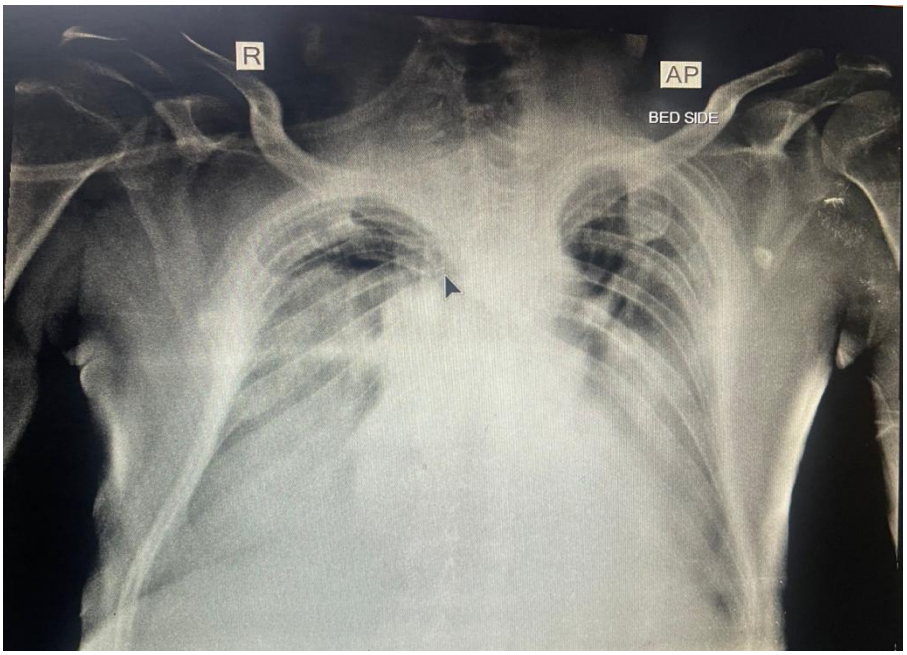
Ongoing research into targeted therapies may offer new treatment options in the future.

DISCUSSION

Achondroplasia requires a comprehensive management approach due to the potential complications such as cardiovascular disease such as calcific aortic stenosis,spinal stenosis, obstructive sleep apnea, and recurrent ear infections. The patient's cognitive development was normal, consistent with typical findings in achondroplasia. Early intervention and regular follow-up are critical to managing potential complications and supporting optimal growth and development.

This case underscores the variability of clinical manifestations in achondroplasia and the importance of considering additional genetic factors in atypical presentations. The presence of calcific aortic stenosis, severe thoracolumbar kyphosis, early-onset osteoarthritis, and hearing impairment necessitates a broader diagnostic approach and individualized management plan.





LOKPRIYA HOSPITAL
SAMRAT PALACE, GARH ROAD, MEERUT - 250003

DEPARTMENT OF NON-INVASIVE CARDIOLOGY

DATE : 01.07.2024 REFERENCE NO. : 7487
PATIENT NAME : SUBHASH CHAND AGE/SEX : 52YRS/M
REFERRED BY : DR. T.R. SIROHI ECHOGENECITY : NORMAL
REFERRING DIAGNOSIS : To rule out structural heart disease.

ECHOCARDIOGRAPHY REPORT

DIMENSIONS		NORMAL			NORMAL
AO (ed)	2.6 cm	(2.1 - 3.7 cm)	IVS (ed)	1.4 cm	(0.6 - 1.2 cm)
LA (es)	2.8 cm	(2.1 - 3.7 cm)	LVPW (ed)	1.3 cm	(0.6 - 1.2 cm)
RVID (ed)	1.2 cm	(1.1 - 2.5 cm)	EF	55%	(62% - 85%)
LVID (ed)	4.0 cm	(3.6 - 5.2 cm)	FS	27%	(28% - 42%)
LVID (es)	2.7 cm	(2.3 - 3.9 cm)			

MORPHOLOGICAL DATA :

Mitral Valve: AML	: Normal	Interatrial septum	: Intact
PML	: Normal	Interventricular Septum	: Intact
Aortic Valve	: Calcific	Pulmonary Artery	: Normal
Tricuspid Valve	: Normal	Aorta	: Normal
Pulmonary Valve	: Normal	Right Atrium	: Normal
Right Ventricle	: Normal	Left Atrium	: Normal
Left Ventricle	: Normal		

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Services : Ambulance Blood Bank 24 घण्टे इमरजेंसी सेवा
Helpline Numbers - 0121- 2792500, 2601901, (M) 8194007414

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:: 2 ::

2-D ECHOCARDIOGRAPHY FINDINGS :

Severe Calcific Aortic Stenosis (PG/MG- 109 /73mmHg), Mild AR. Concentric LVH. LV Diastolic Dysfunction. Adequate LV Systolic Dysfunction (LVEF =55%). Mild MR, Mild TR, Mild PAH.LA/RA are normal in size. RV normal in size with adequate contractions. No intracardiac mass/ clot /vegetation /pericardial effusion.

DOPPLER STUDIES :

Valve	Regurgitation	Velocity m/sec	Gradient mmHg
Mitral Valve	Mild	0.92	3.2
Tricuspid Valve	Mild	1.6	11.0
Pulmonary Valve	No	0.68	1.7
Aortic Valve	Mild	5.2/4.0	109/73

IMPRESSION :

- *Severe Calcific Aortic Stenosis (PG/MG- 109/73mmHg). Mild AR*
- *Concentric LVH.*
- *LV Diastolic Dysfunction.*
- *Adequate LV Systolic Dysfunction (LVEF = 55%).*
- *Mild MR, Mild TR, Mild PAH.*

DR. SANJEEV KUMAR BANSAL
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Lokpriya Heart Centre

DR. HARIOM TYAGI
MD, DM (Cardiology)
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Director, Lokpriya Heart Centre

NOTE: Echocardiography report given is that of the procedure done on that day and needs to be correlated clinically. This is not for medico legal purpose, as patient's identity is not confirmed. No record of this report is kept in the Hospital.

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CONCLUSION

This rare case of achondroplasia with unusual clinical features highlights the complexity of the condition and the necessity for a multidisciplinary approach to management. Further research into genetic modifiers may provide insights into the variability of phenotypic expressions in achondroplasia.

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