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Andersen-Tawil Syndrome (ATS) – A case report

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Abstract: *Andersen–Tawil syndrome (ATS) is rather unique in the family of channelopathies and is characterized by ventricular arrhythmias, dysmorphic features, and periodic paralysis. ATS is a disorder of cardiac repolarization. This patient is a suspected case of Andersen–Tawil syndrome (ATS). The patient referred in the study came with hypokalemic periodic paralysis with dysmorphic facial features and ventricular arrhythmia which has close resemblance to Andersen-Tawil syndrome.*

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I. Introduction

Andersen-Tawil syndrome (ATS) is characterized by periodic paralysis, cardiac arrhythmias, and distinct facial and skeletal features. The majority of patients with ATS (ATS1) have point mutations in the *KCNJ2* gene, which encodes the inward-rectifying potassium channel known as Kir2.1. The skeletal muscle and cardiac symptoms are accounted for, in most cases, by a dominant negative effect of the mutations on potassium channel current, resulting in prolonged depolarization of the action potential. A little above 100 cases of Andersen-Tawil syndrome have been reported worldwide. Andersen Tawil Syndrome is unique channelopathy and represents the first link between cardiac and skeletal muscle excitability. The genetic defect in ATS is not like that in other forms of potassium sensitive periodic paralysis and is distinct from the long QT syndrome locus.

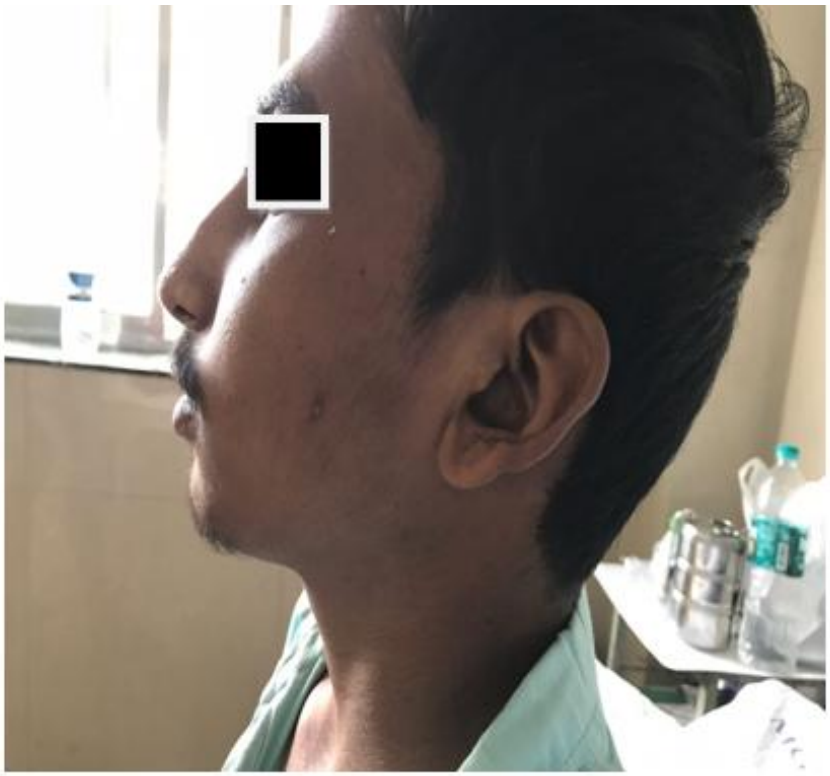


Fig. 1: Showing low set ears, micrognathia and retrognathia

II. Case Report

A 20 year old young boy came with the complaint of weakness in all four limbs which is acute in onset (seizure three episodes), with tongue bite and urinary incontinence. Eyeballs were also up rolled. He was afebrile, pulse was 86/min, blood pressure was 130/80 mmHg, and saturation was 100% on room air. Physical examination showed that the patient has short face.

Systemic Examination:

Central nervous system: Patient had flaccid quadriplegia (secondary to hypokalemic periodic paralysis). Sensory system was normal, facial feature suggestive of low set ears. Other systemic examination was normal.

Ultra-sonography (USG) showed that the right kidney was absent. Further routine investigation showed that the platelets were reduced, potassium was also very less. Confirmed that the Dengue IGM positive. Blood sugar level was 120mg/dl.Treatment for seizure was started and also for reduced potassium level and other problem areas.

Fig. 2: Showing low set ears

Blood examination reports are given below.

REPORTS			
Hb	16.4	14.5	13.6
TLC	17,560	13,130	10,290
DLC	86/9/04	85/12/03	92/04/02
Plt	**0.50**	**0.22**	**0.35**
PCV	47.9	41.90	41.50
Bili T	3.65		0.71

Bili D	1.38		0.22
SGOT	62.0		29.6
SGPT	35.0		45.4
ALKPO4	65		41
TPR	4.88		4.33
Alb	3.04		2.4
BUN	18.7		22.4
Urea	40.0	33.0	48.0
Creat	0.79	0.69	0.70
Uric Acid	2.40		
Na	142.0	144.0	138.0
K	**1.8**	**2.2**	**4.3**
Cl	104.0		
Ca	8.62		

Few other important tests done the results are given below:

- Urine Potassium – 10.3, Urine Spot Potassium – 32.5

His electrocardiograph (ECG) on admission showed ventricular bigeminy with QT interval of 0.36 sec. (Figure 3). Laboratory investigation revealed that serum potassium was 1.8 mEq/L; rest of the investigations including complete blood count, urine routine examination, serum sodium and chloride, serum creatine phosphokinase (CPK), arterial blood gas analysis and thyroid profile were all normal.

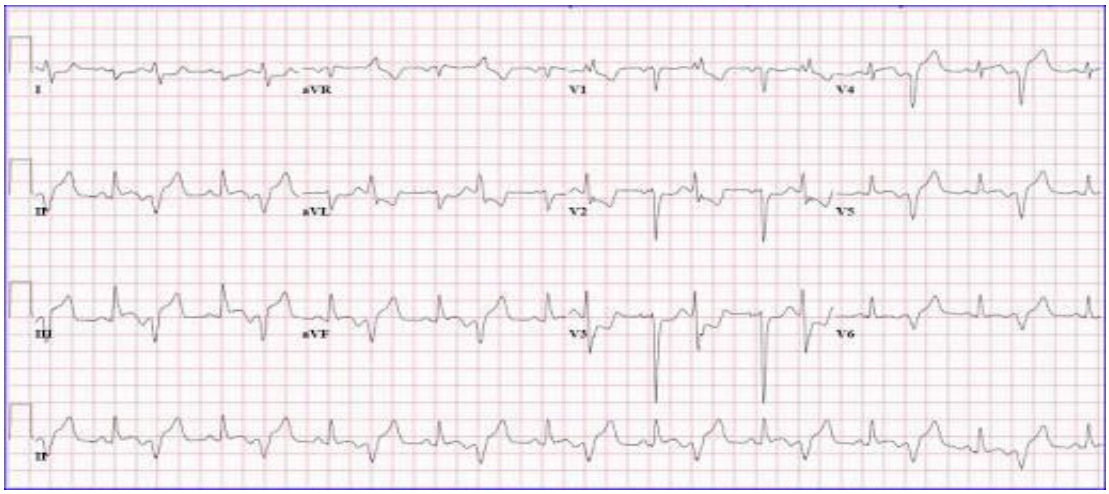
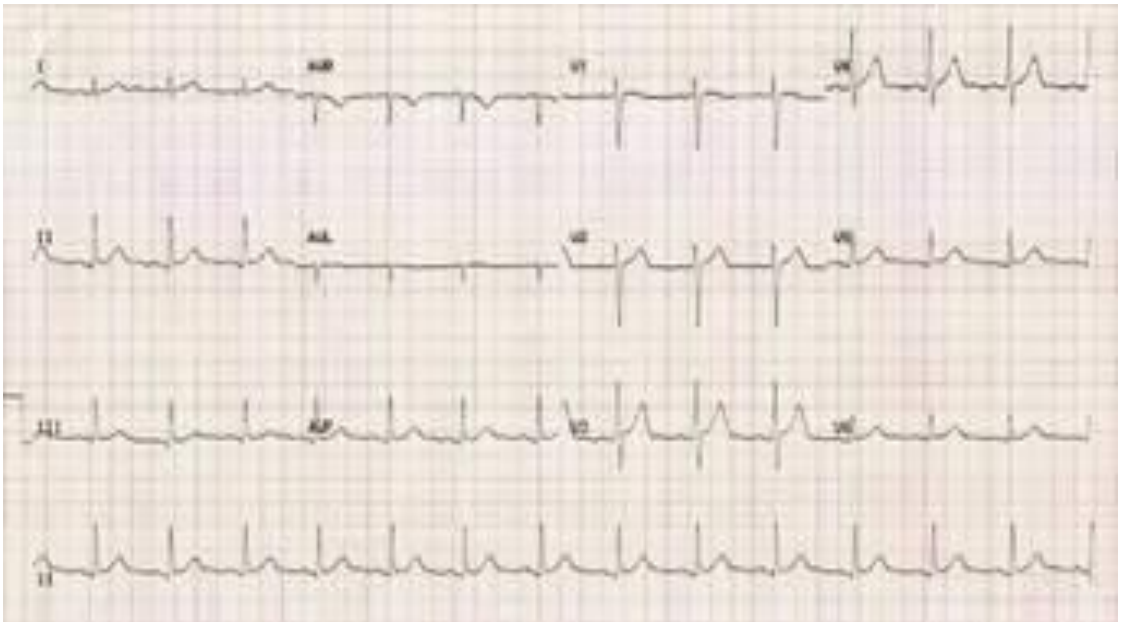


Fig. 3: ECG showing ventricular bigeminy



ECG showing normal sinus rhythm

He was treated with potassium supplementation (initially intravenous and subsequently oral) and on the 2nd day of hospitalization his quadriplegia completely resolved. Repeat serum potassium was 4.3mEq/L and repeat ECG showed normal sinus rhythm. Trans-thoracic echocardiography ruled out any structural heart disease. Nerve conduction study and Electromyography undertaken subsequently were normal.
Based on the findings and physical examination and clinical traid of hypokalamic paralysis, facial dysmorphism and ventricular arrhythmias the patient was labeled as a suspected case of "Andersen-Tawil syndrome. However genetic study would require to confirm the diagnosis.

III. Discussion

Anderson-Tawil Syndrome is an uncommon heterogeneous autosomal dominant disorder characterized by periodic paralysis, dysmorphic features and cardiac arrhythmias. Andersen-Tawil Syndrome (ATS) is a rare potassium channel disorder, characterized by episodic weakness, ventricular arrhythmias and dysmorphic features (short stature, scoliosis, clinodactyly, hypertelorism, small or prominent low set ears, micrognathia and broad forehead). The full spectrum of the disease is characterized by ventricular arrhythmias, dysmorphic features, and periodic paralysis. ATS is a disorder of cardiac repolarization. A little above 100 cases of Andersen-Tawil syndrome have been reported worldwide. Andersen Tawil Syndrome is unique channelopathy and represents the first link between cardiac and skeletal muscle excitability. Donaldson, Yoon, Fu & Ptacek (2009) mentioned that Mutations in *KCNJ2* are the primary cause of Andersen-Tawil Syndrome (ATS) with 21 mutations discovered in 30 families. These mutations affect channel function through heterogeneous mechanisms, including reduced PIP_2 -related channel activation and altered pore function.

IV. Conclusion

Anderson-Tawil Syndrome is very rare disease and also it needs expertise to diagnose the same. The patient in the present study had many resemblances with the signs and symptoms reported in Anderson-Tawil Syndrome. With all the symptoms and physical examinations the case has been labeled as Anderson-Tawil Syndrome. Clinical genetic testing of ATS type 1 is often not possible due to lack of facility and diagnosis of type 2 is essentially clinical.

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