Observation on a case of Jervell and Lange-Neilsen syndrome in an adult

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A case is described of true Jervell and Lange-Neilsen syndrome in an adult. Some relatives of the patient had QT lengthening, syncopal attacks, pigmented naevi on the skin and/or hypoacusia, and/or sudden death.

In a previous paper (Dal Palù and Furlanello, 1967) we discussed a patient who had recurring hyperkinetic syncopal crises triggered by emotional stress.

In this case the electrocardiogram recorded between the crises revealed a peculiar lengthening of the QT interval associated with a very slow repolarization wave and possible fusion of the TU waves, which appeared well defined in the ventricular postextrasystolic complexes.

Comparable anomalies of the electrocardiogram were observed and reported for the first time by Jervell and Lange-Nielsen (1957), then by Levine and Woodworth (1958), Romano, Genne, and Pongiglione (1963), Fraser, Froghatt, and James (1964), Ward (1964), Jervell, Thingstad, and Endsjö (1966), Sánchez Cascos, Sánchez-Harguindeguy, and de Rábago (1969), and Gale et al. (1970). The tracings were obtained in children suffering with syncopal attacks. In many instances, these subjects were congenitally deaf mute or affected by severe auditory deficiencies and often they died quite suddenly of incontrollable ventricular fibrillation. Necropsy did not reveal morphological cardiopathies.

According to James (1967), similar clinical manifestations have been seen in certain strains of Dalmatian dogs. The breeder noticed that these genetically related animals were deaf-mute, presented chromatic anomalies of the coat, and died suddenly. Electrocardiogram showed a uniform lengthening of the QT interval; death was caused by ventricular fibrillation. These observations are thought-provoking because these animals can provide excellent material for comparative genetic studies.

Because of the symptoms and typical pattern of the electrocardiogram, the case presented below may be diagnosed as Jervell and Lange-Nielsen syndrome.

A thorough review of past and current published reports failed to reveal any cases of the syndrome diagnosed in adult life.

Case report

The patient is a 61-year-old man affected from an early age by hearing deficiency (hypoacusia). At the age of 59 he had his first attack of syncope and was in hospital two weeks ago. The patient was admitted recently to his local hospital after a second attack. During this stay, syncope recurred frequently and were related as a cause-effect to ventricular fibrillation, as was shown by the electrocardiogram tracings (Fig. 1). For these reasons the patient was sent to the intensive care unit of the University Hospital where he underwent repeated electrical defibrillation.

Later the patient was transferred to our medical ward for further investigation and management.

The family history and the genetic profile outlined in the pedigree (p. 651) do not include the personal recollections of the parents because of their age.

The study revealed that several members of the family died suddenly during syncopal crises. Meanwhile the incidence of hearing defects and the presence of intradermal naevi were high.

The electrocardiograms obtained from the proband's sibs were characterized by typical lengthening of the QT interval, especially in Innocente's, Giuseppina's, and Maria's electrocardiograms, the latter showing also a slow repolarization wave with terminal fusion of the TU waves (see Fig. 4a–5b). Large intradermal naevi were found...
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**FIG. 1** Patient's electrocardiogram during a syncopal attack. Spontaneous repetitive ventricular fibrillation of short duration is initiated by a ventricular extrasystole during the vulnerable phase.

**FIG. 2 and 3** Patient's electrocardiograms registered at monthly intervals between the crisis. Both are grossly abnormal. Note the length of the T-U waves (freq. 77/m'; Q-TU: 0.57, respectively 60/m' and 0.61).
FIG. 4a, b  Brother and sister of the patient. Both cardiograms are characterized by a lengthening of the QT interval. The brother's cardiogram freq. 60/m', QT O'48, prominent U wave. The sister's freq. 71/m', Q-T O'46, prominent U wave.

FIG. 5a, b  The son and sister of the patient. The son's electrocardiogram is very similar to his father's: QT interval is only slightly prolonged. The sister's is grossly atypical: TU waves are fused. (freq. 45/m', Q-TU O'62.)
on inspection of the skin of some of the patient’s
sibs. The blood and serum studies gave normal results.

On admission the patient appeared anxious but
his general status was satisfactory except for a
moderate arterial hypertension of atherosclerotic
type (180/90 mmHg), with other clinical findings
suggesting sclerosis of the aorta, supported by
radiological evidence. Some naevi were noted on
the skin, and one of them was large, located in the
dorsal area.

The hearing defect was detected by audiometric
evaluation (frequency: 4000 c.p.s.).

The clinical pathological studies of the blood
and serum, including the evaluation of the elec-
trolytic pattern (K+, Na+, Cl-, Ca++), were
normal.

The electrocardiograms shown in Fig. 2 and 3
were obtained at monthly intervals between
the crisis of tachyarrhythmia. They revealed a
conspicuous lengthening of the QT interval (0.58
sec; frequency: 70/m²).

The repolarization waves were abnormal: the
fusion of the terminal phase of the T wave with
the U wave is clear (see D3 and aVF of the
second electrocardiogram).

Moreover, there were signs of interatrial block
and of left ventricular hypertrophy.

The patient, since his first attack, has been
maintained on digoxin 0.25 mg daily. His condi-
tion has improved and the electrocardiogram
shows a decrease of the QT interval.

Discussion

The main clinical features shown by the case
presented here, may be summarized as fol-

(a) Recurring episodes of hyperkinetic
syncopes; (b) pathological lengthening of the
ventricular electrical systoles; (c) abnormal
pattern of the repolarization wave which
cannot be related to an electrolytic imbalance as
revealed by serum studies; (d) high incidence
of sudden death, syncopal attacks, QT
lengthening, hypoaucia, and pigmented naevi
among many relatives of the patients.

As far as ventricular fibrillation is con-
cerned, there is no doubt about its close re-
lation with the abnormal length of the QT
interval which prolongs that vulnerable period
of the cardiac cycle when occasional ectopic
impulses may produce a chaotic repetitive
paroxysmal arrhythmia, as has been proved by
Delore (‘R in T’ syndrome).

The treatment of choice for these functional
cardiopathies — both in theory and as a result
of our experience with this case — appears to
be digitalization, which causes a shortening of
the monophasic wave and therefore of the
QT interval (Olley and Fowler, 1970).

Pathogenesis of the prolonged QT interval
is not clear. Many hypotheses have been ad-
vanced, as an ionic imbalance of the mem-
brane (which is not detectable by standard
techniques of serum studies) or a congenital
metabolic abnormality; both hypotheses may
explain the conspicuous variability of the
shape of the T waves, which is a peculiar
characteristic of all patients with this syn-
drome. Similar QT prolongation and T wave
abnormalities may be produced by asym-
metrical sympathetic stimulation of the ven-
tricles (Yanowitz, Preston, and Abildskov,
1966).

The observation of QT lengthening in
many relatives of our patient, associated with
conspicuous hearing defect and intradermal
naevi, is consistent with true Jervell and
Lange-Nielsen syndrome, even if the pattern
of inheritance is more consistent with Ro-
mano’s and Ward’s syndrome.

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