Principles and Practice of Clinical Electrophysiology of Vision

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Incomplete-Type Congenital Stationary Night Blindness

Yozo Miyake

The Schubert-Bornschein type of congenital stationary night blindness (CSNB) is characterized by a unique, negative-shaped electroretinogram (ERG) in which the amplitude of the a-wave is normal and larger than that of the b-wave.[7] We found that patients with CSNB and a negative-shaped ERG could be subdivided into two types, complete or incomplete, according to rod function.[5] Complete CSNB lacks rod function, but incomplete CSNB shows some rod function on the rod ERG and psychophysical dark adaptation.[2, 5, 8] The cone ERG may be normal to abnormal in either type. Since single–bright-flash ERG had a negative shape in both types (Fig 97–1), we first supposed the incomplete CSNB to be a variant of the complete CSNB, differentiated only by the severity of the functional disturbance. However, our analysis of patients' pedigrees and special functional properties only seen in incomplete CSNB[4, 5] strongly suggested that complete and incomplete CSNB are different clinical entities.[5] Since the cone ERG of incomplete CSNB is abnormal and shows characteristic properties,[4] incomplete CSNB may not be a proper name for this interesting eye disorder.

Recently, Weleber[9] described a patient who had features of Forssius-Eriksson ocular albinism and incomplete stationary night blindness, which raised the issue of whether these two entities are one and the same disease. Further electrophysiological studies in patients with Forssius-Eriksson albinism will answer this question.

The clinical characteristics of incomplete CSNB are described in comparison with complete CSNB and with some similar hereditary disorders.

CLINICAL FINDINGS

Psychophysical Dark Adaptation

In incomplete CSNB, rod adaptation is present, although the final threshold is elevated by approximately 1.0 to 1.5 log units. By contrast, complete CSNB, shows absent rod adaptation.[5]

Visual Acuity and Visual Field

Visual acuity is moderately poor, as in complete CSNB. In our series, corrected visual acuity ranged from 0.1 to 0.8 (mean, 0.41). The visual field was normal with Goldmann V-4 and I-4 targets and was narrower than normal with I-2 targets in most patients.

Refractive Error

The refractive error ranges from mild myopia to hyperopia in many patients. This finding is in marked contrast to that of complete CSNB, where most patients have high to moderate myopia. However, it should be noted that, in our series of incomplete CSNB, a few patients had high myopia.[5] Recently, we found an incomplete-type X-linked pedigree where one patient showed hyperopia (+3 D), one showed emmetropia, and the third showed high myopia (−6.5 D).[3] Since these three patients

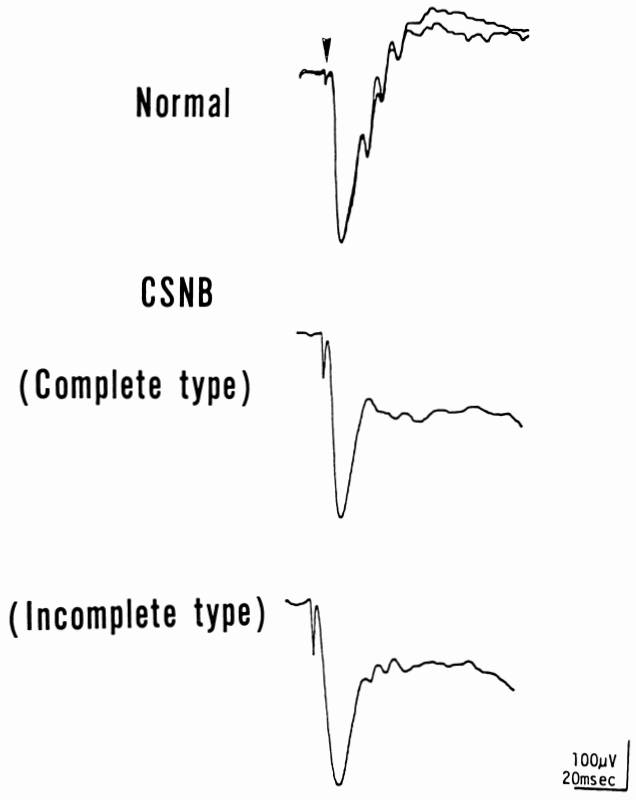

FIG 97–1.
Single–bright-flash ERG after dark-adaptation in complete and incomplete CSNB. The *top* shows a normal control.

had incomplete-type visual function, the possibility of a variation in refractive error among incomplete-type patients is reinforced by the patients in this single family. Such a finding was also reported recently by Khouri et al.[2]

Electroretinography

Single–Bright-Flash Electroretinography

Single–bright-flash ERG has diagnostic power in incomplete as well as complete CSNB. The ratio of b- to a-waves is below 1.0 in all patients, which indicates a negative-shaped ERG (see Fig 97–1). The mean and 2 SD for the ratio of b- to a-waves in incomplete CSNB (0.54 ± 0.20) is larger than in complete CSNB (0.45 ± 0.24), and this has statistical significance ($P < .05$). The oscillatory potentials in incomplete CSNB are more frequently recordable than in complete CSNB. Namely, 20 of 23 patients with incomplete CSNB and 11 of 35 patients with complete CSNB showed detectable oscillatory potentials.

Scotopic (Rod) Electroretinography

The rod ERG is subnormal in incomplete CSNB and absent in complete CSNB (Fig 97–2).

Photopic (Cone) Electroretinography

The cone and 30-Hz flicker ERGs in incomplete CSNB are very small; however, those of complete CSNB are normal to subnormal (see Fig 97–2).

Suppression of 30-Hz Flicker Electretinograms During Dark Adaptation

We found that all patients with incomplete CSNB showed an exaggerated increase in amplitude and a universal characteristic change in waveshape during light adaptation following enough dark adaptation.[4] Figure 97–3 shows changes in the 30-Hz flicker ERG during light adaptation in a patient with incomplete CSNB (left) and changes in relative amplitude in ten patients (right). When recorded after 30 minutes of dark adaptation, the amplitude is extremely low but increases exaggeratedly during light adaptation. In other words, the 30-Hz flicker ERG is unusually suppressed by dark adaptation in incomplete CSNB.

Although an increase in amplitude is also observed during light adaptation in normal subjects and allied diseases (such as complete CSNB, congenital retinoschisis, retinitis pigmentosa, cone dystrophy, or Oguchi's disease),[4] such characteristic changes in amplitude and waveshape have not been seen during light adaptation.[4] This result suggests that an evaluation of the increase in amplitude and waveform during light adaptation may be of potential value in diagnosing incomplete CSNB.

Electro-oculography

The light peak-vs.-dark trough ratio was normal in 11 of 16 patients with incomplete CSNB.

Fundus and Fluorescein Angiography

The fundus appearance is essentially normal. Some patients may show slight temporal optic disc pallor or some goldish metallic reflex in the superior midperipheral retina. The macula is normal, and good foveal reflex is observed (Fig 97–4). Fluorescein angiographic findings are normal even in aged patients (Fig 97–5).

Other Ocular Findings

Nystagmus and/or exotropia may be associated in some patients.

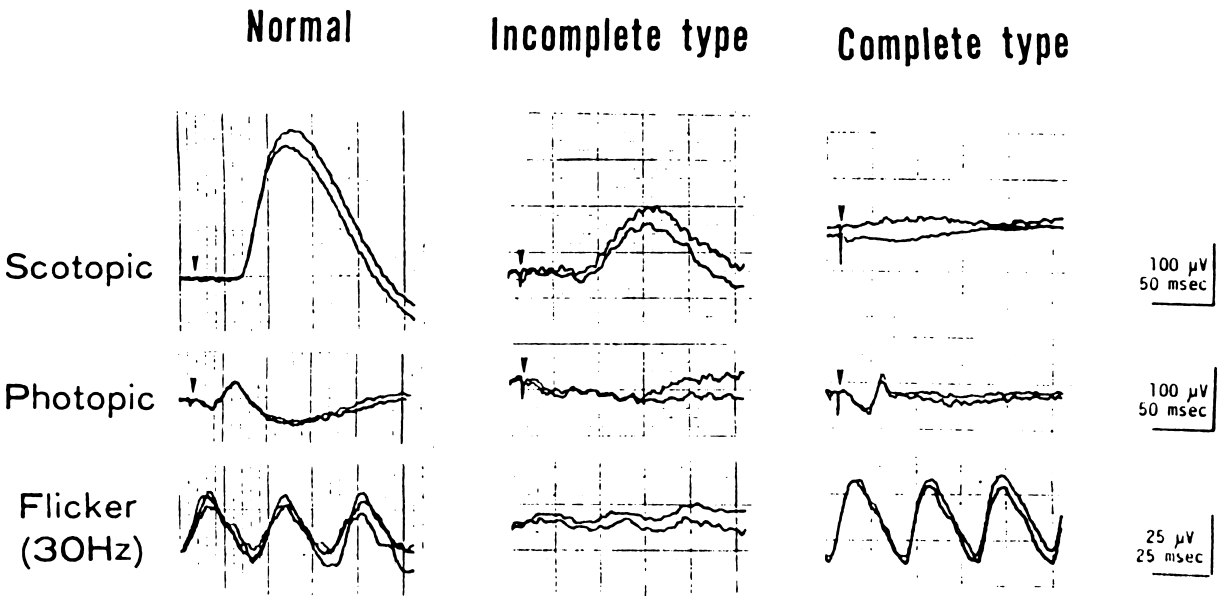

FIG 97–2.
Scotopic (rod), photopic (cone), and 30-Hz flicker ERGs in incomplete and complete CSNB. The *left* ERGs are normal controls.

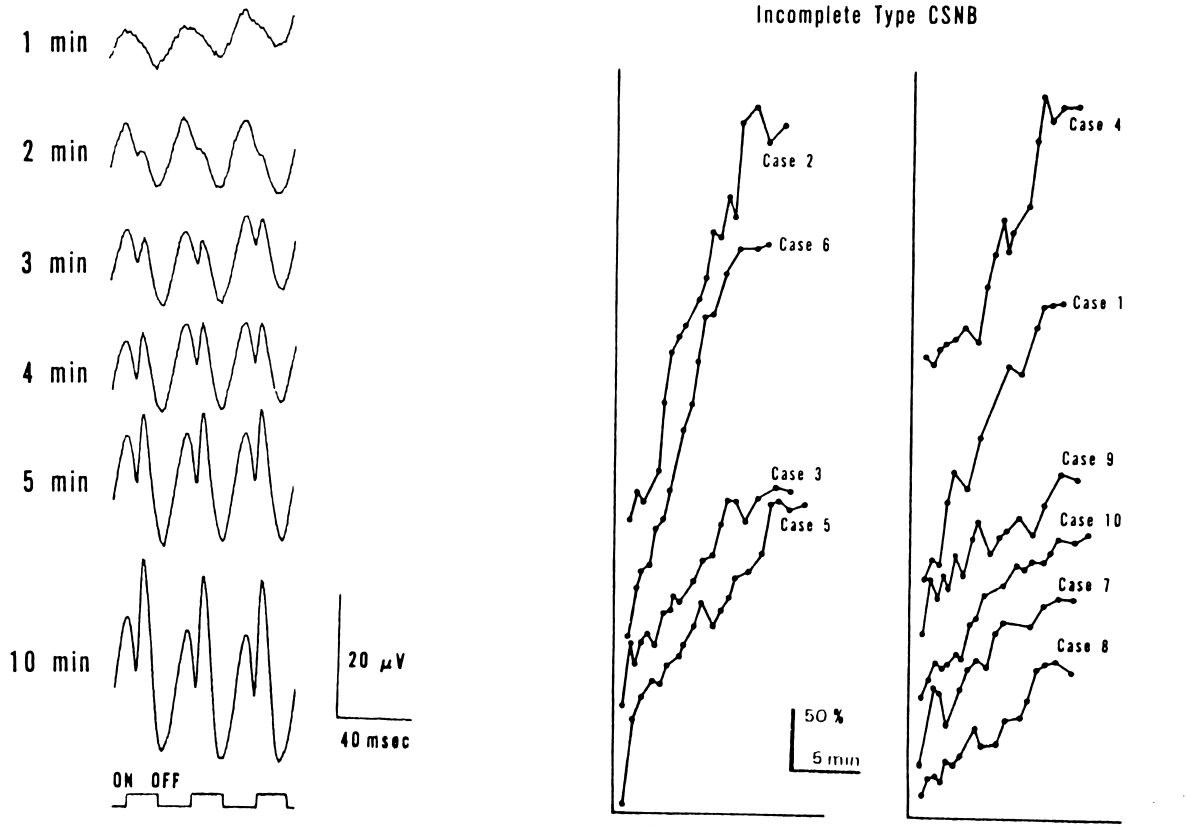

FIG 97–3.
Changes in the 30-Hz flicker ERG during light adaptation in incomplete CSNB *(left)* and changes in relative amplitude in ten patients *(right)*. The curve for each patient is shifted vertically to prevent overlap. The calibration mark indicates the size of a 50% increase in amplitude. (From Miyake Y, Horiguchi M, Ota I, Shiroyama N: *Invest Ophthalmol Vis Sci* 1987; 28:1816–1823. Used by permission.)

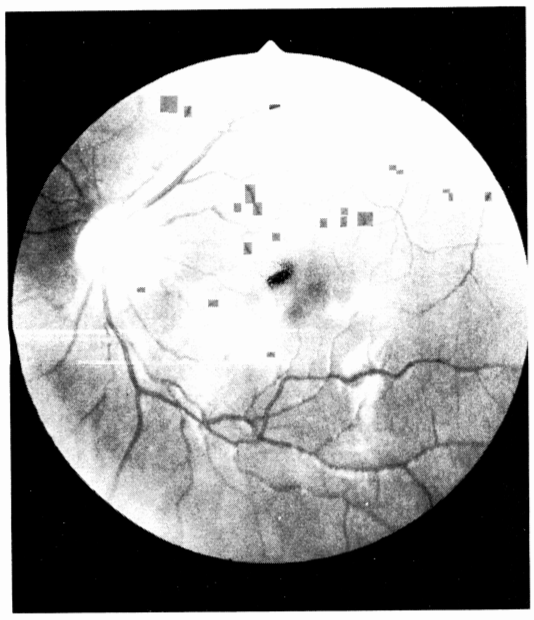

FIG 97–4.
Ophthalmoscopically normal macula in incomplete CSNB.

Hereditary Transmission

In our series of patients with incomplete CSNB, all were male, and five pedigrees were most likely X-linked recessive. Recently, Khouri et al.[6] also reported a family of incomplete CSNB with X-linked recessive transmission.

CHANGES IN VISUAL ACUITIES

Figure 97–6 shows changes in visual acuity during follow-up of incomplete CSNB. Most patients wore correcting lenses and showed little change or a slight improvement in visual acuity during follow-up. These results suggest that refractive error in incomplete CSNB is no different from normal.

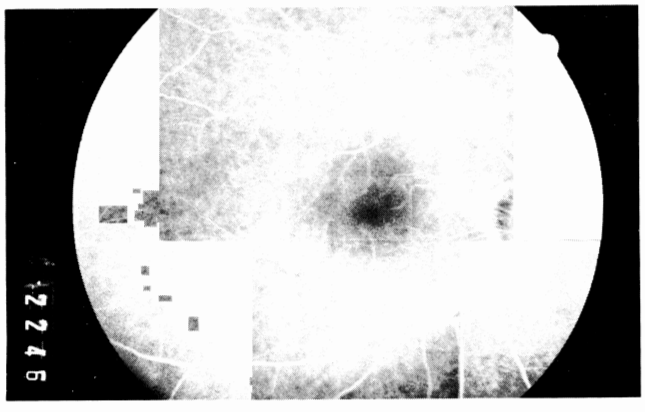

FIG 97–5.
Fluorescein angiography in a 75-year-old patient with incomplete CSNB.

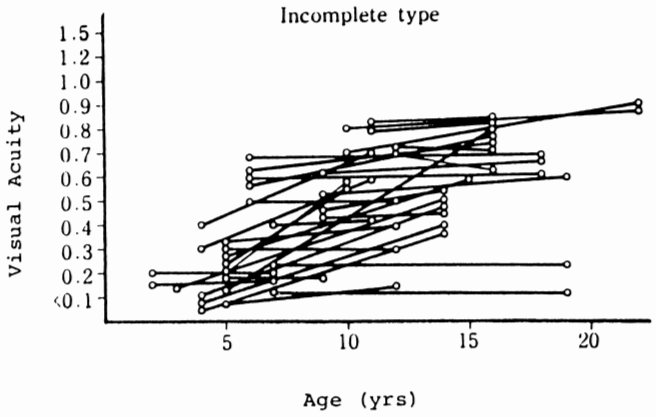

FIG 97–6.
Changes in visual acuity during follow-up of incomplete CSNB.

DIFFERENTIAL DIAGNOSIS

In 1954, Riggs[3] reported three patients with autosomal dominant CSNB who showed an almost complete lack of rod function but perfectly normal cone function; their ERGs had a positive shape. These patients' conditions were clearly different from incomplete CSNB. In 1964, Auerbach et al.[1] reported patients to be of the "Riggs type" since their ERGs had a positive shape. However, unlike the patients reported by Riggs,[6] these patients showed some rod function, and their photopic ERGs were abnormal. The incomplete CSNB is rather similar to the "Riggs type" reported by Auerbach et al.[1] Although patients with incomplete CSNB showed a negative ERG and Auerbach's "Riggs-type" patients showed a positive ERG, this difference may be a result of the different stimulus intensities used by us[5] and Auerbach and associates.[1] If this is the case, our complete and incomplete CSNB may correspond to group I (Schubert-Bornschein type) and group II (Rigg's type), respectively, in Auerbach's classification. We found a definite difference between complete and incomplete CSNB in cone function on the ERG.[5] However, Auerbach et al.[1] reported no such difference in their comparison of groups I and II. Also, unlike our incomplete CSNB, there were female patients in Auerbach's report.[1] Further studies may be needed to differentiate the incomplete CSNB from Auerbach's "Riggs type."

There are some similarities between incomplete

CSNB and X-linked congenital retinoschisis. A hereditary mode, moderately disturbed visual acuity, hyperopic refractive error, a negative ERG, a moderately disturbed rod and cone ERG, and a normal EOG are common findings in both disorders. However, the essentially normal fundus appearance, particularly the normal macular finding, in incomplete CSNB is different from the abnormal macula seen in congenital retinoschisis. The characteristic change in 30-Hz flicker ERG during light adaptation is not seen in congenital retinoschisis.[4] The stationary condition of the incomplete type is seldom seen in congenital retinoschisis.

Finally, it should be noted that incomplete CSNB is easily overlooked or misdiagnosed as established eye diseases such as amblyopia, optic atrophy, or tapetoretinal dystrophy. ERG analysis can provide a correct diagnosis of this newly indentified rod-cone dysfunction syndrome.

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