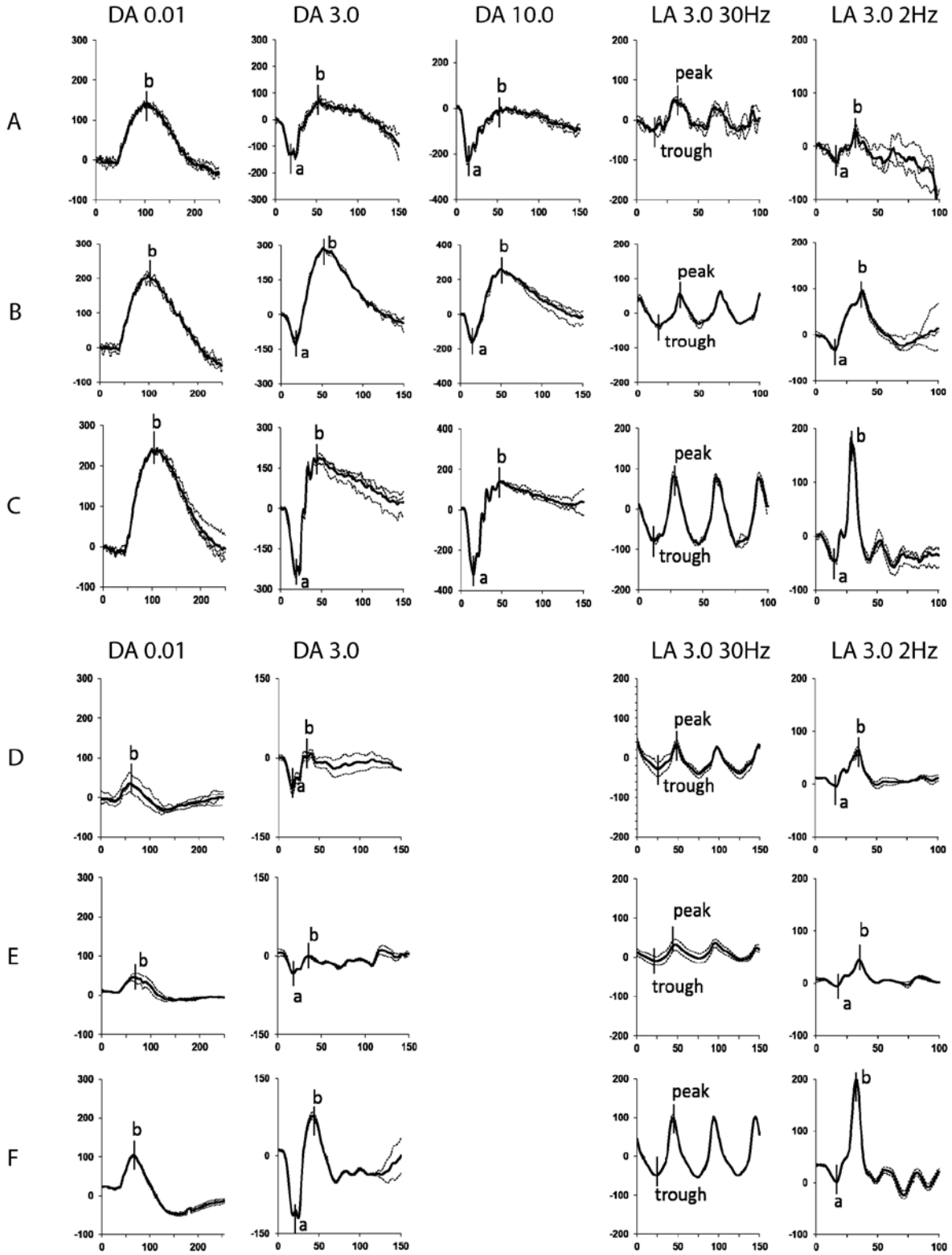
Figure e-1: Full-field electroretinogram findings in Lafora disease



A-F: Represent full-field electroretinogram (ERG) traces from patients and controls. Dark adapted (DA) and light adapted (LA) testing were done to delineate rod and cone system function, respectively. Stimulus names are abbreviated in the accepted nomenclature; this includes adaptive state of the eye (DA or LA) followed by stimulus intensity in $cd.s.m^{-2}$. Under DA conditions, up to three different intensity stimulus were used (0.01, 3.0 and 10.0 $cd.s.m^{-2}$). Under LA conditions two different stimuli was used at 3.0 $cd.s.m^{-2}$ (30 Hz and 2Hz). The recordings were done at two centers (*EPM2A* patients in Toronto and *EPM2B* patients in Genova) using two machines, and hence, two control traces are shown.

A: ERG traces from Patient-1. The DA 10.0 shows normal a-wave indicative of normal rod photoreceptor function; but b-wave is markedly reduced consistent with selective generalized rod bipolar cell dysfunction (reduced b/a ratio). The LA ERGs show reduced amplitudes indicative of generalized cone system dysfunction (cone photoreceptors and bipolar cells).

B: ERG traces from Patient-2. All DA responses (DA 0.01, DA 3.0 and DA 10.0) were normal. LA ERGs show delayed peak times indicative of generalized cone system dysfunction.

C: Control ERG trace using Diagnosys LLC system in Toronto.

D: ERG traces from Patient-3. The DA responses show evidence of generalized reduction in rod photoreceptor (reduced a-wave of DA 3.0) and rod bipolar cell function (reduced b-wave of DA 3.0; reduced b/a ratio). The LA ERGs show moderately reduced amplitudes indicative of generalized cone system dysfunction.

E: ERG traces from Patient-4: The DA responses show similar abnormalities as seen in case 3. The LA ERGs show moderately reduced amplitudes indicative of generalized cone system dysfunction.

F: Control ERG trace using CSO-Retimax system in Genova.