INDIANA UNIVERSITY
November 13,1998

Gregory Smith, M.D.
8937 Southpointe Drive, #2C
Indianapolis, IN 46227
Re: Nathan
Dear Dr. Smith,
Nathan was last seen November 12, 1998. At that time he underwent an examination under anesthesia and performance of an electroretinograrn for possible retinal dystrophy.
Examination: Intraocular pressures were normal as was the anterior segment slit lamp exam. Dilated fundus examination was notable for attenuation of the retinal vessels diminished foveal light reflex, a reticulated paflern of the mid- peripheral retinal pigment epithelium. The optic nenes appeared normal.
Electroretinogram was markedly abnormal under anesthesia. The scotopic or rod responses were essentiallv flat. The photopic or cone responses were significant for markedly diminished amplitudes. Some of this depression is undoubtedly due to the anesthesia, however, the ERG remains markedly abnormal despite the confounding variable.
The abnormal ERG and the findings present on detailed fundus examination are both consistent with a retinal dystrophy Possible etiologies are extremely diverse.
Differential diagnosis includes:

1) Retinitis pigmentosa and retinitis pigmentosa associated diseases. Inheritance pafferns are quite variable. Chromosome testing is possible for some varieties. Nathan does not have other findings consistent with either Usher syndrome or Bardet-Biedl syndrome.
2) Leber's congenital amaurosis but this is unlikely given Nathan's apparently normal acuity and the absence of nystagmus.
3) Congenital stationary night blindness. This is decreased night vision with either normal or mild to moderately reduced central visual acuity. This is a possible diagnosis however these patients are usually myopic whereas Nathan is hyperopic.
4) Other multi-system diseases such as infantile phytanic acid storage disease and infantile neuronal ceroid lipofuscinosis. Hallmark of these storage diseases is progressive neurologic abnormalities.

Recommendations:
I agree with the planned comprehensive neurologic evaluation. My second recommendation is evaluation by the genetics department. Chromosome analysis can be pefformed to possibly idenufy a rhodopsin gene mutation, as found in some cases of retinitis pigmentosa.. A significant portion of our evaluation will be observation over time. Without a pedigree of other affected family members, it is difficult to provide prognostic information. We would like to see Nathan in our office again in four months. Please feel free to contact our office if we can be of additional support in his evaluation. We appreciate the opportunity to participate in the care of this child.

Sincerely,

Daniel E Neely M. D.
Assistant Professor of Ophthalmology
DEN/sd

Cc: Angela Tomlin, PhD
Bhuwan Garg, M.D.
To The Parents

The following test results are very hard to read.
The first page are the notes that were taken during Nathan's ERG that DR. Neely annotated. The following pages are the actual output from the ERG. The lines during night testing are suppose to look like a normal bell curve, Nathan's results are fairly flat. The day light results are supposed go up and down symmetrically. Nathan's day light results are up and down very erratically. I am more than happy to forward copies to anyone who might be interested, or you could contact Dr. Neely at Riley Children's Hospital (I only included 2 pages of ERG output test results because of server space restrictions).

Notes Taken While the ERG was being Perfomed

7022012
C-016
(FEB 95)

Clarian Health
Methodist · IU · Riley

ROTARY EYE

SPECIAL EYE-RECORD

PEDIATRIC OPHTHALMOLOGY
(Pre-op Notes)

4 yo [illegible] [illegible] in dark
[illegible] OD/OS

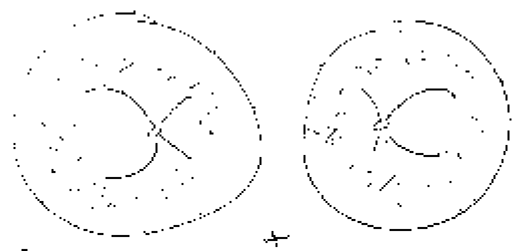
MILTO, NATHAN
14344438

☐ In-Patient ☐ Out-Patient

Signed [illegible] Date: NOV 1 2 1998

(Operative Notes)

[illegible]



① normal nerve [illegible] macula
Signed Date: NOV 1 2 1998
② Mild calculated mid peripheral pigment changes. No Bone spicules.
③ moderate vessel attenuation

(Postoperative Notes)

ERG ① Scotopic responses: severely diminished - flat
② Photopic responses: moderately depressed

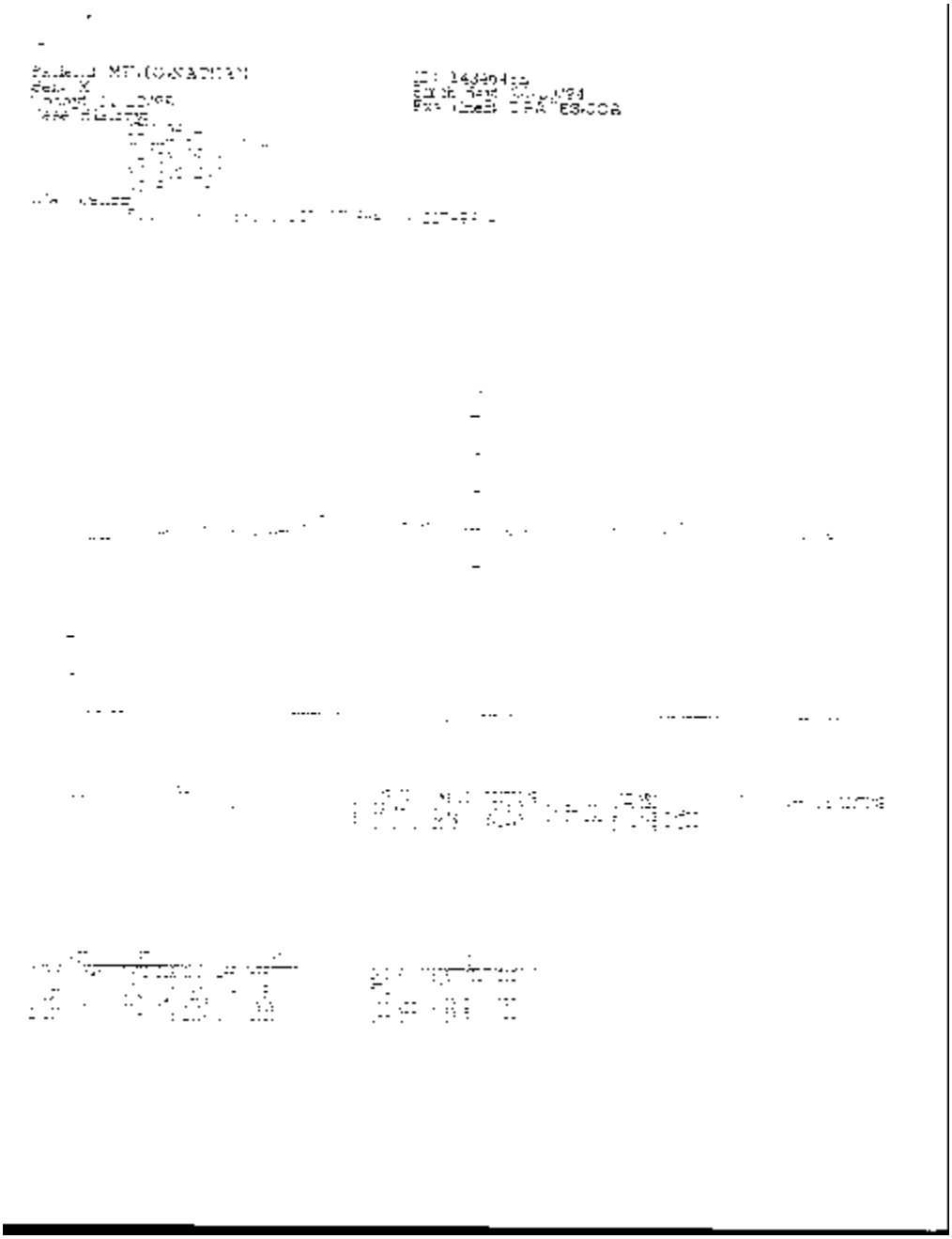
Signed: [illegible] Date: NOV 1 2 1998

Medical Record Copy

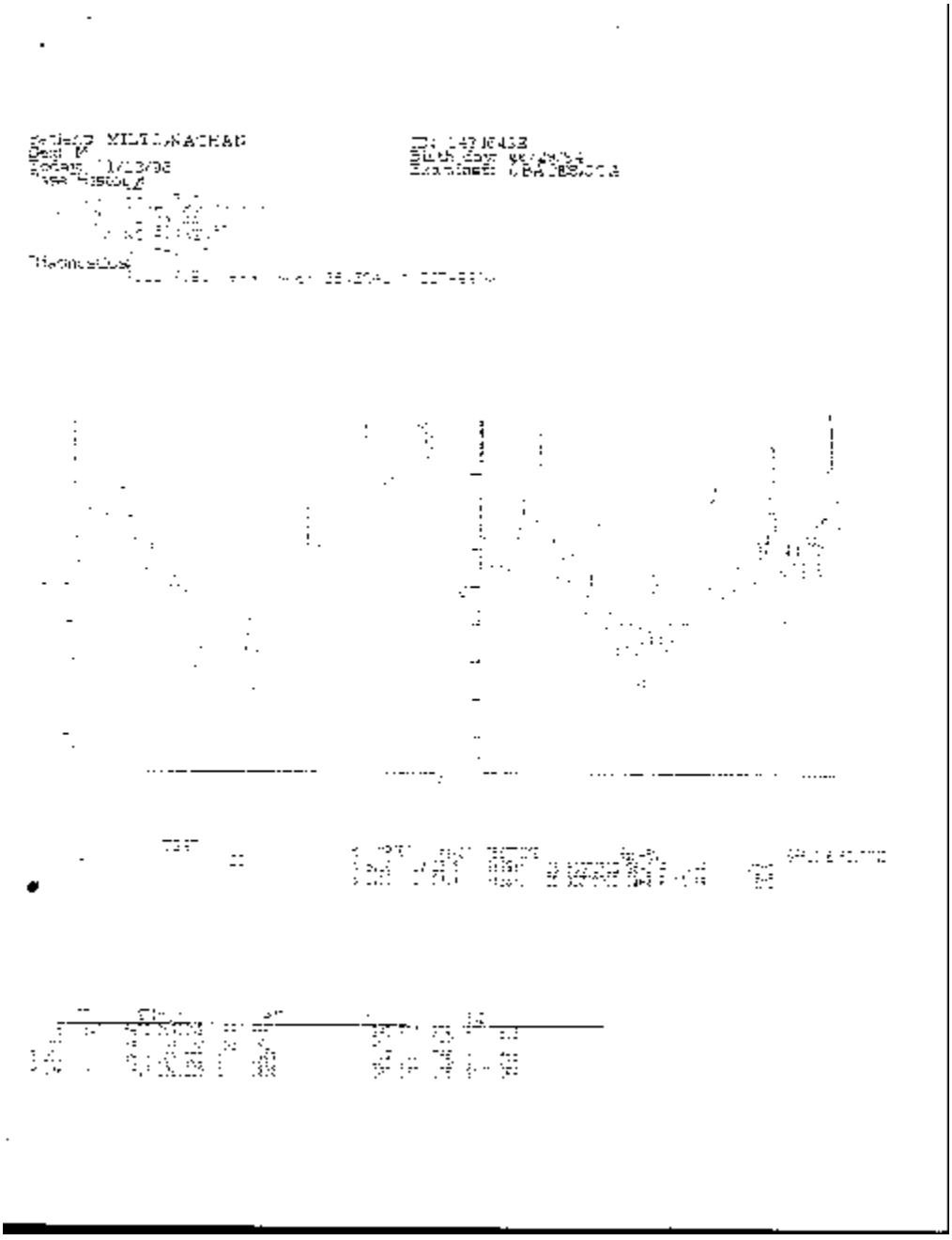
SPECIAL EYE RECORD **B-70**

E-CLIN. NOTES	L-LAB	C-X-RAY	D-DIAGNOSTIC	M-SURGERY	Q-THERAPY	T-ORDERS	W-NURSING	Y-MISC.

ERG Results Page 1 Night Light Testing



ERG Results Page 2 Day Light Testing



ERG RESULTS #3 DAY LIGHT TESTING (not shown, available upon request)

ERG TEST #4 RESULTS NIGHT LIGHT (not shown, available upon request)

ERG RESUTLS #5 NIGHT LIGHT (not shown, available upon request)

ERG RESULTS #6 NIGHT LIGHT TEST (not shown, available upon request)