

Ophthalmology Clinic Note 625

VONTUNGELN, RACHEL - 425-37-28

* Final Report *

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*** Final Report ***

We had the pleasure of meeting Rachel and her parents for the first time today. She was referred to us by Dr. Picker, Genetics. She has also seen Dr. Mukai, retina service, at Massachusetts Eye and Ear Infirmary. The concern is that she has a retinal degeneration and possible optic neuritis. Another diagnosis that has been considered by other physicians is chronic chorioretinitis. Rachel is a nine years old.

She has had decreased vision for years. Her mom described difficulties suggestive of night vision problems that have been noticed in the last year. She trips over things and bumps into things and they wondered about the visual field. There have been long-term problems with learning and behavior. There is evidence of regression; her mom cites regression of an IQ from 80 to 60. She has been seen by a neurologist in the past, outside Children's Hospital; the last examination by a neurologist was approximately a year ago.

There is also a history of longstanding headaches. There possibly are some delays in development. Episodes of "meltdowns" are mentioned. Possibly there are some autistic features.

Because of inattention, an MRI and CT was done in 05/2008. We are told that showed normal brain. More recently there was an evaluation in Developmental Medicine (06/26/2009). The report of that evaluation is not yet available.

Because of the concern about vision and possible chorioretinitis, in about May of this year she was seen by Dr. Mukai, retina specialist at the Eye and Ear. Cells in the vitreous, optic nerve pallor, attenuated retinal vessels and possible early bull's-eye change and peripheral pigment epithelial changes were noted. ERG testing was suggested, but was not done.

There is some history that at about age five to seven years an examination by an optometrist indicated that vision was at an acceptable range although acuities were poorer than 20/20.

We thank her parents for completing the yellow history sheets. We have reviewed those. There are no eye diseases in the family. She has three younger siblings. Julie and London were slow to talk but now their development is on schedule. The eyes and vision of all three siblings have seemed fine.

Rachel did grade 1 two times. She was in second grade this past year. She has lots of modifications and accommodations in school; there are persistent questions about her performance and school placement going forward.

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EXAMINATION:

Her glasses were read as +1.25 -0.75 X 170 and +1.25 -0.75 X 007.

It was possible to get a measure of acuity using isolated Lea symbols. The result with binocular viewing and also for each eye alone was 20/250. Dr. Baglieri did the acuity testing. Inconsistent responses were noted.

The subsequent cycloplegic retinoscopy today was essentially the same as she is wearing, +1.00 -0.75 X 180, both eyes.

Her eyes were straight at distance and near. No nystagmus was seen. Ductions were full and versions comitant.

On external examination no abnormalities were seen. The pupils and their reactions appeared normal.

At the slit lamp, the structures in the anterior segments appeared normal, both eyes. After the pupils had been dilated, we saw approximately 1+ cells, both eyes. The intraocular pressures were judged to be normal to palpation, both eyes.

As for visual fields, on confrontation with a small toy we had the impression that the visual field is constricted. However, her attention during this evaluation was intermittent.

We attempted measurement of her color vision but could not complete that testing.

On indirect ophthalmoscopy, the media were clear, both eyes. The optic nerve heads are quite pink, flat and have sharp margins. There does appear to be some temporal pallor that is about the same in extent and intensity right eye and left eye. In the macula, the foveal reflexes are diminished and there is irregular pigmentation there. There is at the center of the macula, a darker appearance and the irregular pigment surrounding that does give overall a lighter annulus and thus suggestive of a bull's-eye configuration. The retinal arterioles are mild to moderately attenuated. In the extramacular fundi there is some mild granularity with faint clumping of pigment. Fundus photographs have been taken. The photos have been reviewed and are informative; the photos are to document her current condition and to follow it over time.

SUMMARY AND RECOMMENDATIONS:

This examination shows evidence of retinal degeneration with a prominent maculopathy. Visual acuity is low, both eyes; it is at a legal blind level. There are night vision complaints. Confrontation testing suggests constriction of visual field.

There is a longstanding history of learning problems and concern about regression more recently. Thus, we would be concerned about a neurodegenerative disorder.

Next steps will be those which will lead to a specific diagnosis and those that will provide support for her vision and other needs.

Today we have completed the request for registration with the Massachusetts Commission for the Blind. This is to be faxed to the Commission.

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We did think that it would be well to characterize her vision further by measurement of her dark-adapted visual threshold and Goldmann visual fields. I have suggested that we aim to complete that within the next one to two months. We would also like to perform examination under anesthesia including electroretinography. We are looking tentatively at a September date for that.

We are pleased that she is scheduled to see Dr. Darras, Neurology, 07/07/2009. She will also be returning to Dr. Picker, Genetics, in 08/2009.

We also recommend a low vision consult with Dr. Kathy Miller in perhaps a month or so.

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Completed Action List:

- * Perform by FULTON MD, ANNE B on July 02, 2009 09:20 EDT
- * Sign by FULTON MD, ANNE B on July 02, 2009 15:25 EDT July 02, 2009 15:25 EDT
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