CASE RECORDS of the MASSACHUSETTS GENERAL HOSPITAL



# Case 25-2024: A 12-Year-Old Boy with Autism and Decreased Vision

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## PRESENTATION OF CASE

*Dr. Amy E. Armstrong-Javors:* A 12-year-old boy with autism was admitted to this hospital because of decreased vision in both eyes.

The patient had been in his usual state of health until 6 weeks before this admission, when he began to have difficulty seeing in both eyes. His vision was decreased when he woke up in the morning, improved throughout the day, and worsened again at night. Three weeks before the current admission, the patient told his parents about the vision changes. They took him to an optometry clinic for evaluation. Eyeglasses were not prescribed; a routine annual follow-up evaluation was scheduled.

During the next 3 weeks, the patient noticed that the decrease in vision worsened. He typically walked on his toes and held onto his parents as he walked; however, 4 days before this admission, his parents noticed that he leaned on them heavily while walking. Two days before the current admission, they observed him walking into doors and walls and bumping into objects. On the day of this admission, the patient woke up screaming and panicked because he could not see. His parents brought him to the emergency department of this hospital.

On evaluation, the patient reported that his vision had improved slightly throughout the day. He described darkening of his vision that was worse in the left eye than in the right eye. He was having difficulty identifying movement and details of objects, although he was able to see shapes and colors. The parents reported that the patient had been having puffiness and crusting of the eyes for 2 days, with no eye redness or pain. Review of systems was notable for 3 days of diarrhea and nausea. He had no fever, headache, rashes, mouth sores, joint pain, or weakness.

The patient was born prematurely, at 7 months' gestation. Labor and delivery had taken place outside the hospital setting, and perinatal hypoxia had occurred. The patient had been taken urgently to another hospital, where he was resuscitated and then hospitalized in the neonatal intensive care unit for 2 months. The patient had autism and attention deficit–hyperactivity disorder (ADHD). He had

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developmental delays in speech, language, cognition, and fine motor skills. He had received all routine childhood vaccinations and was taking risperidone; there were no known drug allergies.

The patient lived with his parents and older sister, all of whom were healthy. Ten months before this admission, they had moved from an urban area to a rural area in New England. The patient was in sixth grade and was enrolled in special education classes at school. He had been an avid player of video and virtual reality games, but his parents had removed the gaming systems from their home 2 months before the patient's vision changes had begun. He continued to watch videos on the computer and television; his parents noted that he did not sit close to the devices and sometimes looked at the wall instead of the screen. The patient's parents described him as a "picky eater." His diet consisted almost exclusively of hamburgers, french fries, ranch dressing, glazed doughnuts, and juice boxes. The patient avoided trying new foods and did not take vitamin pills or gummies because he disliked the taste or texture.

**Table 1. Laboratory Data.***

| Variable | Reference Range† | On Admission |
|---|---|---|
| White-cell count (per μl) | 4500–13,500 | 5700 |
| Hemoglobin (g/dl) | 13.0–16.0 | 12.3 |
| Hematocrit (%) | 37.0–49.0 | 37.0 |
| Mean corpuscular volume (g/dl) | 78.0–98.0 | 89.4 |
| Platelet count (per μl) | 150,000–450,000 | 216,000 |
| Sodium (mmol/liter) | 135–145 | 137 |
| Potassium (mmol/liter) | 3.4–5.0 | 4.1 |
| Chloride (mmol/liter) | 98–108 | 104 |
| Carbon dioxide (mmol/liter) | 23–32 | 20 |
| Urea nitrogen (mg/dl) | 5–20 | 6 |
| Creatinine (mg/dl) | 0.60–1.50 | 0.38 |
| Glucose (mg/dl) | 70–110 | 77 |
| Aspartate aminotransferase (U/liter) | 10–40 | 17 |
| Alanine aminotransferase (U/liter) | 10–55 | 20 |
| Alkaline phosphatase (U/liter) | 129–417 | 422 |
| Total bilirubin (mg/dl) | 0.0–1.0 | 0.4 |
| Albumin (g/dl) | 3.3–5.0 | 3.6 |
| Total protein (g/dl) | 6.0–8.3 | 5.9 |
| Prothrombin time (sec) | 11.5–14.5 | 17.1 |
| Prothrombin-time international normalized ratio | 0.9–1.1 | 1.4 |

* To convert the values for urea nitrogen to millimoles per liter, multiply by 0.357. To convert the values for creatinine to micromoles per liter, multiply by 88.4. To convert the values for glucose to millimoles per liter, multiply by 0.05551. To convert the values for bilirubin to micromoles per liter, multiply by 17.1.

† Reference values are affected by many variables, including the patient population and the laboratory methods used. The ranges used at Massachusetts General Hospital are for children who do not have medical conditions that could affect the results. They may therefore not be appropriate for all patients.

On examination, the temporal temperature was 36.2°C, the blood pressure 104/71 mm Hg, the heart rate 94 beats per minute, and the oxygen saturation 98% while the patient was breathing ambient air. The height was 156 cm (70th percentile), the weight 40.3 kg (38th percentile), and the body-mass index (BMI; the weight in kilograms divided by the square of the height in meters) 16.6. The patient was pale, alert, interactive, and wary of the physical examination.

*Dr. Ryan A. Gise:* Mild periorbital swelling was present. The visual acuity in both eyes was limited to hand motion. The pupils were symmetric and reactive to light, and the extraocular movements were full. Color-vision testing, automated perimetry, fundus photography, and optical coherence tomography could not be performed because the patient had poor vision and was unable to cooperate. A slit-lamp examination was notable only for keratinization of the conjunctiva in both eyes. A funduscopic examination revealed pallor of the optic disk in both eyes. Examination of the peripheral retina was limited because the patient was unable to cooperate.

*Dr. Armstrong-Javors:* Strength, sensation, reflexes, and coordination were normal. A gait examination revealed toe walking; the patient asked for assistance when walking. The abdomen was nontender. Horizontal ridges were present on the toenails (Fig. 1), and there were scattered bruises on the arms and legs. No ulcers or rashes were seen. Laboratory test results are shown in Table 1. The patient was admitted to this hospital.

*Dr. Camilo Jaimes:* During the next 2 days, imaging was attempted but limited because the patient was anxious about the testing. On the third hospital day, magnetic resonance imaging (MRI) of the head and orbits, with and without the intravenous administration of contrast material, was successfully performed while the patient was under sedation (Fig. 2). A fluid-sensitive short-tau inversion recovery (STIR) sequence showed

subtle hyperintensity of the mid-intraorbital segments of the optic nerves in both eyes, with no enhancement. The optic chiasm, optic tracts, and optic-nerve sheath complexes appeared normal. Diffuse heterogeneous thickening of the calvarium was present.

*Dr. Armstrong-Javors:* Diagnostic tests were performed.

## DIFFERENTIAL DIAGNOSIS

*Dr. Eric D. Gaier:* This 12-year-old boy with autism presented with a 6-week history of progressive vision loss. Careful attention to the patient's description of the vision changes may provide clues to the diagnosis in this case. "Darkening" of the vision is not typical of corneal, lenticular, or cortical processes, but it is a common descriptor used for disorders of the anterior visual pathway (retina and optic nerve). Visual acuity that is limited to hand motion suggests profound vision loss involving the peripheral retina, which could not be examined in this patient. The preservation of brisk reactivity of the pupils suggests some contribution of an outer retinal process, a functional overlay, or both. The presence of nonspecific optic-disk pallor on funduscopic examination and a subtle hyperintense signal of the optic nerves on fluid-sensitive STIR MRI suggests a primary optic neuropathy but does not rule out a retinal process. I will construct a differential diagnosis focusing on causes of optic neuropathy, with particular consideration of this child's risk factors, which are based on his history.

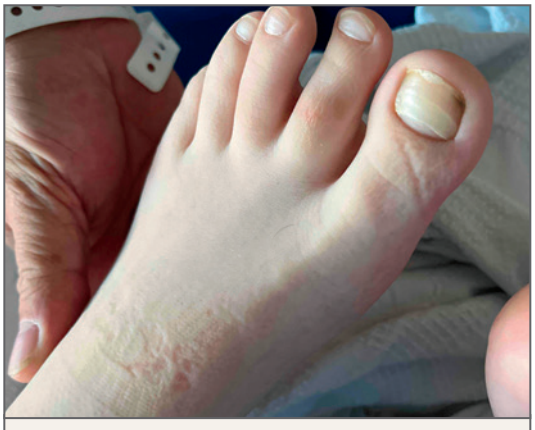


**Figure 1. Clinical Photograph of the Toenails.**
A photograph obtained on the day of the current admission shows horizontal ridges on the toenails.

### OPTIC NEUROPATHY

Causes of optic neuropathy can be divided into conditions associated with optic-disk swelling and those associated with optic-disk atrophy, although it is important to acknowledge that abnormal swelling can evolve to atrophy. The optic-disk pallor observed in this patient is indicative of atrophy but does not rule out previous swelling. With few exceptions, the appearance of the optic nerve should not be used to discern the underlying cause of optic neuropathy because of considerable variation within and overlap between causes.

There are many causes of optic neuropathy (Fig. 3). Prematurity and neonatal hypoxia can be associated with optic atrophy. However, such a process would have resulted in nonprogressive vision impairment that would have been recognized much earlier, rather than developing at 12 years of age. The patient had no history of trauma or exposure to toxins or radiation. He had no systemic symptoms or uveitis that would be suggestive of infection. Compressive lesions were reliably ruled out on neuroimaging. Vascular causes of optic neuropathy are rare in children without systemic risk factors. Infiltration of the optic nerve by cancer manifests with progressive swelling, not atrophy.

### AUTOIMMUNE OR INFLAMMATORY DISEASES

Types of autoimmune optic neuropathy — also known as optic neuritis — include demyelinating disease associated with multiple sclerosis, neuromyelitis optica spectrum disorder (NMOSD), and myelin oligodendrocyte glycoprotein antibody disease (MOGAD). Both NMOSD and MOGAD can cause profound bilateral vision loss, and MOGAD has accounted for a large portion of cases of optic neuritis in children since it was discovered and testing became available.[1] Inflammatory optic neuropathy is less common than optic neuritis and can occur in the context of a systemic disease such as sarcoidosis or antineutrophilic cytoplasmic antibody–associated disease. Both optic neuritis and inflammatory optic neuropathy are unlikely in this patient because the onset of vision loss was slower than would be expected with these conditions, eye pain and optic-disk edema (findings typical of

MOGAD) were absent, and there was no optic-nerve enhancement on MRI. Furthermore, he did not have signs or symptoms that would suggest a systemic autoimmune or inflammatory disease.

### GENETIC DISEASES

The patient's parents reported that he had a tendency to look at the wall instead of the screen when using the computer or television. This finding, known as eccentric viewing, is characteristic of the central vision loss that occurs with genetic, toxic, or nutritional optic neuropathy.

The most common type of genetic optic neuropathy is autosomal dominant optic atrophy, which is caused by variants in *OPA1*, a gene that encodes a ubiquitously expressed protein essential for mitochondrial function. Autosomal

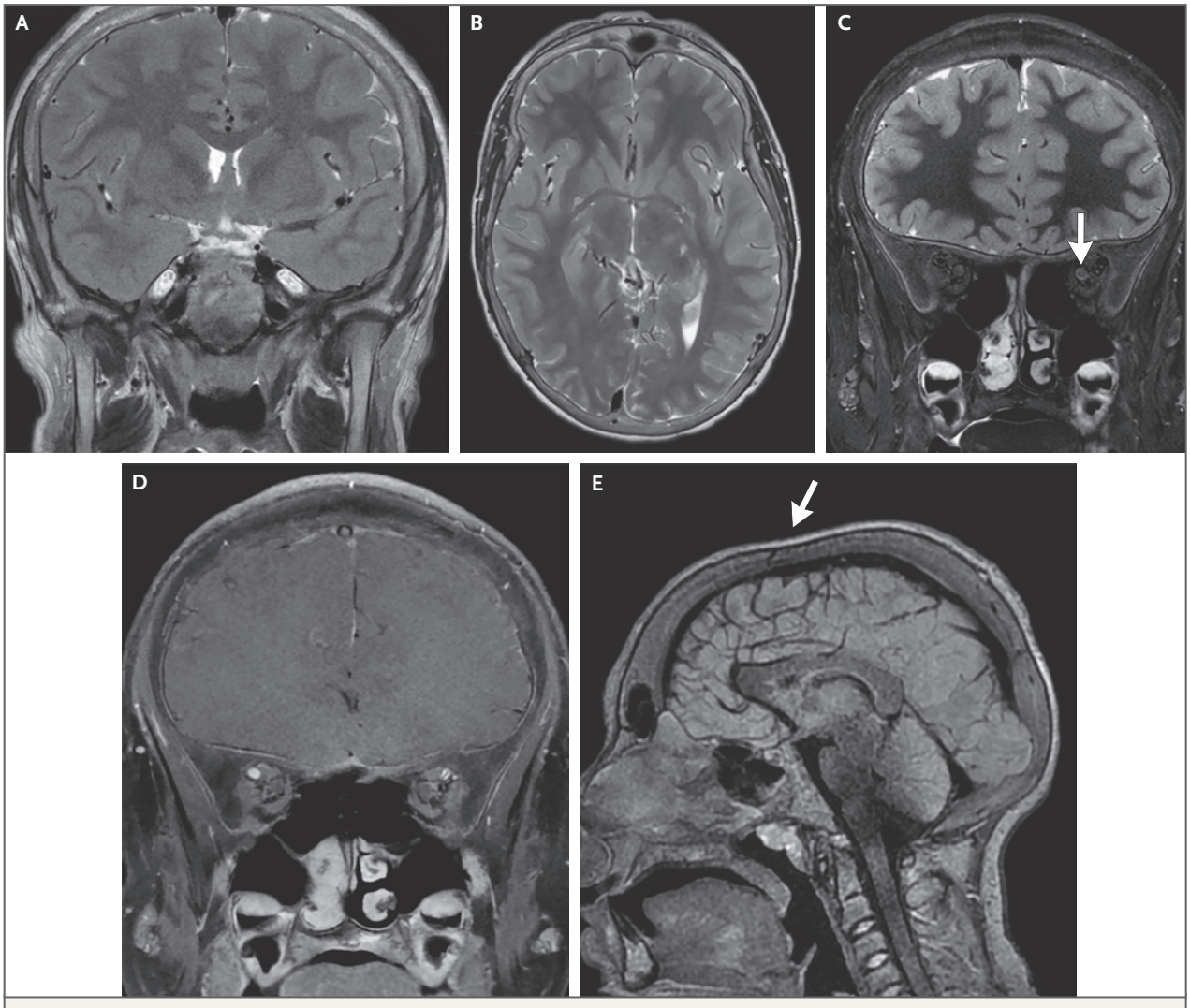



**Figure 2. MRI of the Head and Orbits.**

A coronal T2-weighted image of the head and optic chiasm (Panel A) and an axial T2-weighted image of the optic radiations (Panel B) show no abnormalities. A high-resolution coronal image from a fluid-sensitive short-tau inversion recovery sequence of the orbits (Panel C) shows a very subtle hyperintense signal in the central portion of the optic nerves (arrow), which is more conspicuous in the left eye than in the right eye. A high-resolution coronal T2-weighted fat-suppressed image of the orbits (Panel D) shows no abnormal enhancement in the optic nerves or elsewhere in the orbit. A sagittal three-dimensional image from a fluid-attenuated inversion recovery sequence (Panel E) shows marked thickening of the calvarium (arrow), without focal lesions; the intracranial contents appear normal.

dominant optic atrophy is characterized by a slow, insidious loss of central vision in both eyes. Other types of syndromic optic atrophy that are caused by genetic variants usually have a similarly slow progression or have an early onset of symptoms. Leber's hereditary optic neuropathy, which is caused by variants in the mitochondrial gene *ND4*, results in acute-to-subacute vision loss that often occurs in one eye weeks before it occurs in the other. Leber's hereditary optic neuropathy is maternally inherited. The absence of a family history does not rule out a genetic cause because of variable expressivity and incomplete penetrance.[2] However, the tempo of vision loss in this patient was more rapid than the pattern seen with autosomal dominant optic atrophy; Leber's hereditary optic neuropathy is uncommon and is associated with less severe impairment of visual acuity than that observed in this patient, but it remains a possibility.

#### TOXINS

Many elements and compounds from common sources can cause toxic optic neuropathy. The onset of vision loss may not be rapid if exposure is low but consistent, with the concentration of the toxin in the body building over a long period of time. This patient had not taken any medications associated with toxic optic neuropathy (e.g., linezolid, ethambutol, or halogenated hydroxyquinolines) or eaten any contaminated foods associated with this condition.[3] Exposure to methanol or ethylene glycol would be expected to cause anion gap metabolic acidosis, which was not present in this patient. The recent move to a rural area, which had occurred just before the onset of symptoms, could have led to exposure to new toxins, such as those in well water (lead, mercury, cobalt, arsenic, organic solvents, and pesticides).[4] In the absence of identified exposures, profound neurologic deficits, and prominent gastrointestinal manifestations, toxic optic neuropathy seems unlikely.

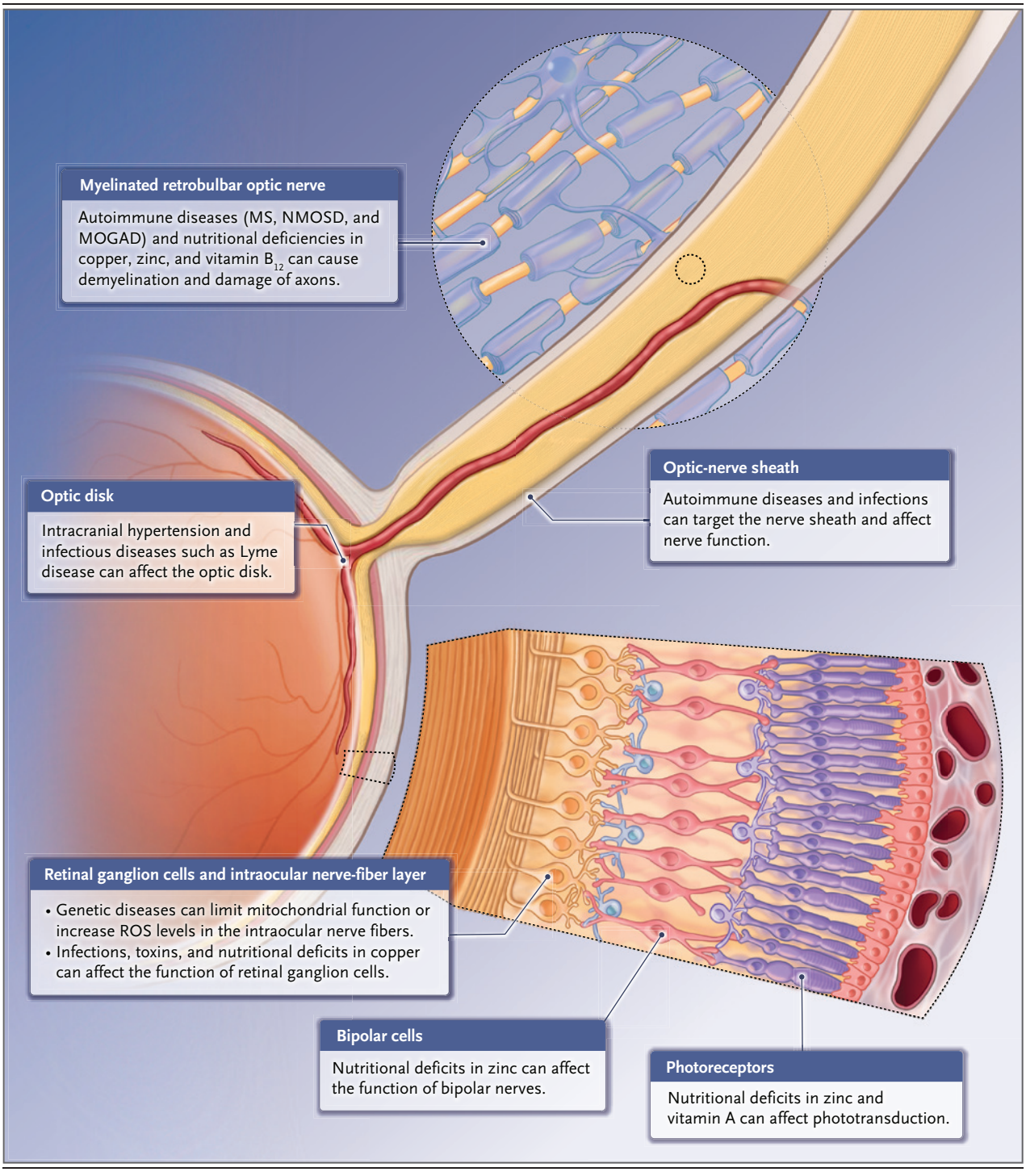
#### NUTRITIONAL DEFICIENCIES

Nutritional optic neuropathy and retinopathy manifest with symmetric painless vision loss that is gradual and progressive. Central vision is often affected in patients with nutritional optic neuropathy, as in those with genetic or toxic optic neuropathy, which reflects a shared mitochondrial pathogenesis.[5] Although nutritional deficiency is more likely to occur in resource-limited countries because of malnourishment, this patient's symptoms are consistent with nutritional optic neuropathy. Selective eating is common in patients with autism and has resulted in nutritional deficiencies associated with disorders affecting the optic nerve and retina.[6]

Vitamin A includes fat-soluble compounds that are essential for the chemical cycle that supports phototransduction in retinal photoreceptors. Patients with vitamin A deficiency can have decreased sensitivity to light in dark conditions, known as nyctalopia, which was present in this patient. Vitamin A deficiency may also be associated with pigmentary changes in the peripheral retina, xerophthalmia (dry eyes), and characteristic squamous metaplasia (keratinization) of the conjunctiva, called Bitot's spots. Such changes in this patient would be highly suggestive of vitamin A deficiency. The heterogeneous signal in the calvarium on this patient's MRI may reflect hyperostosis, which has been reported in conjunction with vitamin A deficiency in some cases and may cause compressive optic neuropathy when it affects the optic canal.[7] Finally, the elevated international normalized ratio seen in this patient could indicate concurrent deficiency of another fat-soluble vitamin (i.e., vitamin K), which suggests that multiple nutrients may be deficient.

Vitamin $B_{12}$ deficiency has also been reported to cause optic neuropathy in patients with autism and selective eating.[8] However, the normal sensation to light touch, intact deep-tendon reflexes, and absence of anemia make vitamin $B_{12}$ deficiency unlikely in this patient.

Deficiencies in essential trace elements such as copper and zinc can cause optic atrophy and demyelination through dysfunction of the enzymes for which these elements serve as cofactors.[9,10] Copper and zinc are also essential for retinal function.[11] Although copper and zinc deficiencies have been described primarily in the context of bariatric surgery, they can also result from selective eating or a concurrent malabsorptive disorder. The patient's difficulty with walking could reflect myelopathy associated with copper deficiency; the horizontal nail ridges could reflect the Beau's lines associated with zinc deficiency.[12]




I think that optic neuropathy and probable retinopathy associated with nutritional deficiency is the most likely diagnosis. There is especially strong evidence to support the diagnosis of deficiencies in vitamin A, copper, and zinc. Because multiple deficiencies can occur in patients with autism and selective eating,[6] I would recommend broad testing for nutrient deficiencies.

**Figure 3 (facing page). Causes of Optic Neuropathy According to Site of Injury.**

Optic neuropathy can result from processes involving the retina, optic disk, or retrobulbar optic nerve. There are multiple possible causes, and some can occur simultaneously. Autoimmune optic neuropathy — such as multiple sclerosis (MS)–associated optic neuritis, neuromyelitis optica spectrum disorder (NMOSD), or myelin oligodendrocyte glycoprotein antibody disease (MOGAD) — largely involves the myelinated axons of retinal ganglion cells but may also manifest with optic-disk edema. Autoimmune diseases and infections can affect the retrobulbar optic-nerve sheath, causing changes in optic-nerve function. Infections can also affect the optic disk, the axonal portion of the optic nerve, and the retina (including the retinal ganglion cells in the inner retina). Genetic diseases can limit mitochondrial function or cause increased levels of damaging reactive oxygen species (ROS), affecting the particularly susceptible intraocular unmyelinated nerve-fiber layer. Toxins associated with optic neuropathy also target the metabolically susceptible retinal ganglion cells. The sites of injury from nutritional optic neuropathy and retinopathy reflect the various essential roles of vitamins and nutrients, including those involved in phototransduction (vitamin A and zinc), metabolic function (copper and zinc), and structural maintenance of myelin (copper). Causes of optic neuropathy that are not shown include elevated intracranial pressure (with many possible underlying causes), trauma, radiation exposure, vascular disease, and compressive lesions affecting the retrobulbar optic nerve, optic chiasm, and optic tract.

## DR. ERIC D. GAIER'S DIAGNOSIS

Nutritional optic neuropathy due to vitamin A, copper, and zinc deficiencies.

## DIAGNOSTIC TESTING

*Dr. Armstrong-Javors:* In the workup for optic neuropathy, serologic and cerebrospinal fluid testing — performed to rule out infections and autoimmune causes, such as NMOSD and MOGAD — was negative. Neurologic testing was not pursued for evaluation of the abnormal gait. The toe walking with resultant ankle spasticity was chronic, and the patient's need for assistance while walking was probably related to vision impairment, not new gait instability. Nutritional deficiency was deemed to be the most likely cause of the vision changes, given that the patient had a severe case of selective eating and was not taking a multivitamin or supplemental nutrients. Blood tests revealed deficiencies in vitamin A (<5.0 μg per deciliter; reference range, 12.8 to 81.2), copper (46 μg per deciliter; reference range, 75 to 145), and zinc (61 μg per deciliter; reference range, 66 to 110). The patient also had severe deficiencies in 25-hydroxyvitamin D and vitamin C.

## LABORATORY DIAGNOSIS

Nutritional optic neuropathy due to multiple nutritional deficits, including vitamin A, copper, and zinc deficiencies.

## DISCUSSION OF VISION LOSS IN CHILDREN

*Dr. Gise:* In a review of vision impairment in 1393 children in Israel, more than half the cases were attributed to inherited eye diseases, and approximately 8% were attributed to optic atrophy, either inherited or acquired.[13] When the patients with optic atrophy had severe vision impairment (as in this case), another disease process, such as hydrocephalus, was the underlying cause. In a study of optic atrophy in 218 children in the United States, the most common cause was a tumor (in 29%), and only 2 patients (1%) had toxic or metabolic disease.[14] In a more recent study of optic atrophy in 272 children from the same population, the most common cause was prematurity (in 16%), followed by a tumor (in 15%).[15] No cases were attributed to nutritional deficiency. It is clear that nutritional optic neuropathy is rare in modern times in nations with access to basic nutrients.

Vision abnormalities are more common in children with autism than in those without autism. In one study, optic neuropathy occurred in 1.1% of the patients with autism but in only 0.3% of those without autism.[16] The occurrence of optic neuropathy in patients with autism is probably underestimated, given that symptoms may be underreported and examinations may be limited, as in this case.

Many cases of vision loss in children are preventable and reversible. Nutritional optic neuropathy should be treated with aggressive supplementation of deficient vitamins and minerals to maximize the likelihood of vision recovery.

## MANAGEMENT OF SELECTIVE EATING IN CHILDREN

*Dr. Sarah M. Kadzielski:* Up to 70% of children with autism have mealtime challenges.[17,18] Certain sensory factors (such as a strong preference for taste, color, smell, texture, or packaging) and behavioral factors (such as a reliance on routines and rituals) may contribute to selective eating. Furthermore, limiting the diet can be a coping mechanism for anxiety. Selective eating may be reinforced by a hyperactive gag reflex that can lead to oral aversions, by mealtime rituals and habits, or by a natural decrease in the frequency that caregivers serve nonpreferred foods out of a desire to avoid triggering a tantrum or wasting food that the child does not eat.[19-25]

Although sensory and behavioral factors are prominent drivers of selective eating in children with autism, underreporting of symptoms is a concern, and therefore, other causes should be considered. Selective eating might be due to pain or discomfort, medical conditions (e.g., oropharyngeal dysphagia, eosinophilic esophagitis, gastroesophageal reflux disease, celiac disease, or decreased appetite), dental conditions, food sensitivities or allergies, or psychiatric conditions such as eating disorders.

Children with autism and selective eating may meet the criteria for avoidant–restrictive food intake disorder (ARFID), outlined in the *Diagnostic and Statistical Manual of Mental Disorders*, fifth edition. ARFID, which is characterized by an eating or feeding disturbance that leads to a persistent inability to meet nutritional needs, has many overlapping features with neurodevelopmental, psychiatric, and somatic diagnoses and can occur concurrently with these conditions.[26-28] Up to half of children who receive a diagnosis of ARFID may also have autism.[28] This patient most likely meets the criteria for ARFID with nutritional deficiency and a need for nutritional supplementation, and he should be referred for formal psychiatric evaluation and management.

### MEDICAL MANAGEMENT

The medical management of selective eating in children depends on the severity of the condition.[29,30] All cases of selective eating (often referred to as "picky eating") need close monitoring and conservative management strategies, but severe cases (known as "problem feeding") may need diagnostic testing and more intensive strategies. Severe cases of selective eating cause personal or family stress, affect daily activities, or lead to medical complications related to weight or nutritional intake. Patients with selective eating are at risk for underweight, overweight, or obese status.[29] It is important to measure and monitor trends in the height, weight, and BMI, but these are not the only factors to be assessed when determining severity. For example, this patient had normal growth measurements and trends despite his profound nutritional deficiencies.

The initial goal in the management of selective eating is to ensure adequate intake of calories, macronutrients (carbohydrates, fat, and protein), and micronutrients (vitamins and minerals). It is important to obtain a thorough dietary history. In children with autism, the assessment of nutrient levels may be limited by behaviors such as refusal of blood draws. Use of a multivitamin is essential in severe cases of selective eating and is strongly advised in all cases. A trial-and-error approach may be necessary to determine a multivitamin formulation that the patient will accept, such as powdered, chewable, gummy, liquid, or capsule supplements. For patients with weight loss or poor weight gain, intake of high-calorie foods, additional nutritional supplements, or a medication for appetite stimulation may be needed.

### BEHAVIORAL STRATEGIES

When selective eating occurs in children with autism, sensory and behavioral factors often contribute to the condition, even if medical or psychiatric causes have been identified. In this case, the patient's parents were counseled about behavioral modification strategies that can be implemented at home or school.[31,32] One strategy is to follow meal and snack schedules that are regular and controlled, with set times and durations, given that children are unlikely to eat a substantial portion beyond a 30-minute window. Grazing should be avoided because it can interrupt natural patterns of feeling hungry and full. Another strategy is to create a supportive environment, offering an appropriate table and seating in a comfortable location with minimal distractions and with pleasant conversation to model the social aspects of eating. Begging, nagging, and force-feeding should be avoided because these approaches can

create a power struggle, giving the child an opportunity to say "no" to food or forming negative associations with foods and mealtimes. Exposure to situations in which food is involved but the child is not expected to eat — such as shopping for groceries, preparing food, or serving food to others — may be helpful.

When new foods are introduced to children with autism and selective eating, one or two preferred foods can be presented along with the new food. If the child shows resistance to the new food, it can be offered on a separate plate. In addition, food can be presented in a way that appeals to children, such as arranged as an animal face, cut into shapes, or sorted by color. Positive food-related behaviors should be praised, and a reward such as bubbles or stickers can be given. Negative behaviors, such as spitting, throwing food, or food refusal, should be ignored. To prevent or extinguish brand-specific food preferences, foods can be removed from their original packaging and served from a plate, bowl, or plastic bag to limit any association with the packaging.

If conservative behavioral strategies are not effective in broadening the patient's diet for the treatment of nutritional deficiencies, more intensive therapy may be warranted. Feeding therapy that involves consultation with a speech–language pathologist and an occupational therapist often follows a sequential–oral–sensory approach, which addresses sensory factors and gradually exposes children to foods.[33] Another helpful technique is food chaining, which involves making small, incremental changes to preferred foods until a new food has been completely added to the diet. It is important for caregivers to understand that each step in feeding therapy may take substantial time and require repetitive efforts before any progress is seen. An increase in the variety of foods eaten can be harder to achieve than an increase in the quantity of food eaten.[34] Selective eating often decreases gradually with age.[35] Psychologists can address underlying anxiety with cognitive therapy or relaxation techniques, although the results vary, depending on the developmental level of the child.

## FOLLOW-UP

*Dr. Kadzielski:* While the patient was in the hospital, he received supplements of vitamins A, C, D, and K as well as calcium, thiamine, copper, and zinc. A multivitamin with iron, a high dose of vitamin D, and zinc was prescribed at discharge. His nutrient levels eventually normalized.

As a result of behavioral modification strategies implemented by his parents after counseling, the patient has started to eat lettuce and cheese on his hamburgers. A clear, juicelike supplement was added to his daily juice boxes; however, he has recently started to refuse these. He has been referred for outpatient feeding therapy with occupational therapy and applied behavior analysis. Although his selective eating is clearly associated with sensory and behavioral factors, esophagogastroduodenoscopy is planned to rule out other causes.

*Dr. Gise:* Unfortunately, the patient's optic atrophy was severe. His visual acuity in both eyes remains limited to hand motion. This severe degree of vision loss cannot be reversed when it is found at such an advanced stage. If it is found earlier in the disease course, reversing the nutritional deficit can lead to some improvements in vision. Outcomes are variable and very dependent on the time at which the nutritional optic neuropathy is found and the type of inciting deficit.[36]

The patient has been registered with the Massachusetts Commission for the Blind. His parents are working with this organization and his school to obtain low-vision services.

## FINAL DIAGNOSIS

Nutritional optic neuropathy due to multiple nutritional deficits, including vitamin A, copper, and zinc deficiencies.

This case was presented at Pediatric Grand Rounds.

Disclosure forms provided by the authors are available with the full text of this article at NEJM.org.

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