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Two cases of paediatric vitamin A deficiency

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PURPOSE: The human body cannot synthesize vitamin A, making dietary intake essential. Vitamin A plays a key role in maintaining vision, preserving the integrity of epithelial and mucosal surfaces, supporting humoral and cellular immunity, and promoting growth and development.
In developed countries, deficiency among children is rare and mainly associated with chronic gastrointestinal diseases (malabsorption, liver diseases). Very rarely, it results from severely restricted diets.
Hypovitaminosis A affects the ocular surface and impairs the function of both rods and cones, and in rare cases can also damage the optic nerve. If left untreated, it can lead to permanent blindness. Early diagnosis is thus essential.
METHODS: A presentation of two cases of children with hypovitaminosis A treated at the Eye Clinic and the Pediatric Clinic in Ljubljana.
RESULTS: Case 1: 8-year-old girl underwent liver transplantation at age 7 due to neonatal sclerosing cholangitis. She had complained of poor night vision even prior to transplantation. Examination revealed good visual acuity, however electrophysiological tests showed signs of rod system dysfunction and reduced cone function. Vitamin A levels were low. After oral vitamin A supplementation, she is now asymptomatic; rod function is normal and cone function remains slightly reduced.
Case 2: 5-year-old boy presented with tearing, photophobia, and swollen eyelids. He also had difficulties navigating in dark environments. Examination revealed a keratinized cornea with Bitot's spots and punctate epithelial corneal defects. The optic nerve discs were pale; electrophysiological testing later confirmed dysfunction of both rods and cones as well as optic nerve dysfunction. Due to his generally poor condition and fever, he was referred to the Pediatric Clinic, where Pseudomonas aeruginosa sepsis was diagnosed, his vitamin A levels were immeasurable. Brain MRI was normal. Extensive diagnostics ruled out diseases causing malabsorption or disrupted vitamin A metabolism. The deficiency was most likely due to a severely restricted diet. After oral supplementation, his general condition as well as the condition of the ocular surface and retina improved, but optic nerve damage remained. The boy continues to have reduced visual function and is classified as visually impaired.
CONCLUSION: Hypovitaminosis A is extremely rare in children in developed countries; however, its recognition is essential. With timely and appropriate supplementation, permanent vision impairment can be prevented, and normal development of a child is ensured.

Pomanjkanje vitamina A pri otroku – predstavitev dveh primerov

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NAMEN: Vitamina A naše telo ni sposobno sintetizirati, zato je nujno, da ga vnesemo s hrano. Ključno vlogo ima pri ohranjanju vida, ohranjanju celovitosti epitelija in sluznic v telesu, humoralni in celični imunosti ter spodbujanju rasti in razvoja.
Pomanjkanje je v razvitem svetu pri otrocih redko, pojavlja se predvsem pri kroničnih gastrointestinalnih obolenjih (malabsorbcija, jetrne bolezni), zelo redko pa je posledica slabega vnosa zaradi močno omejenih diet.
Hipovitaminoza A povzroča prizadetost očesne površine, okvarjeno funkcijo paličnic in tudi čepnic, redko pa tudi okvaro vidnega živca. Nezdravljena vodi v trajno slepoto, zato je zgodnja prepoznava ključna.
METODE: Predstavitev dveh primerov otrok s hipovitaminozo A, zdravljenih na Očesni in Pediatrični kliniki v Ljubljani.
REZULTATI: Primer 1: 8-deklica je imela zaradi neonatalnega sklerozantnega holangitisa v starosti 7 let transplantacijo jeter. Že pred transplantacijo je tožila o slabem nočnem vidu. Ob pregledu je bila vidna ostrina dobra, elektrofiziološke preiskave so pokazale okvarjeno delovanje sistema paličnic ter znižano delovanje čepnic. Nivoji vitamina A so bili znižani. Po peroralnem nadomeščanju vitamina je sedaj asimptomatska, delovanje sistema paličnic je normalno, čepnic pa še mejno znižano.

Primer 2: 5-letni deček je prezentiral s solzenjem, fotofobijo in oteklimi vekami. Slabše se je orientiral v temnem prostoru. Ob pregledu je imel keratinizirano roženico z Bitotovimi pegami, vidni so bili pikčasti defekti epitela roženice. Na očesnem ozadju sta izstopali bledejši papili, elektrofiziološko smo kasneje potrdili odstopanja funkcije paličnic in čepnic ter motnje prevajanja po vidnem živcu oziroma vidni poti. Zaradi splošnega slabega stanja in povišane telesne temperature je bil napoten na Pediatrično kliniko. Ugotovljena je bila sepsa povzročena s Pseudomonas aeruginoso, vrednosti vitamina A so bile nemerljive, MRI slikanje glave ni pokazalo posebnosti. Obsežna diagnostika je izključila bolezni, ki povzročajo slabo absorbcijo ali moten metabolizem vitamina A. Pomanjkanje je bilo najverjetneje posledica zelo omejene prehrane. Po peroralnem nadomeščanju se je splošno stanje, stanje očesne površine in mrežnice popravilo, ostala pa je prizadetost vidnega živca. Deček ima slabo vidno funkcijo in je kategoriziran kot slaboviden otrok.
ZAKLJUČEK: Pomanjkanje vitamina A je v razvitem svetu pri otrocih redko, vendar je prepoznavanje ključno, saj z ustreznim in pravočasnim nadomeščanjem preprečimo trajno okvaro vida in omogočimo normalen otrokov razvoj.