



Ocular effects of Hypophosphatasia (HPP)

Hypophosphatasia is a rare genetic disease that often affects the development of bones and teeth. Signs and symptoms vary widely and can range from mild to severe. People with HPP might, for example, have frequent bone fractures, skeletal deformities, short stature, and/or premature loss of baby teeth.

One of the less-recognized outcomes of HPP is its effects on the eyes. Ocular manifestations have only recently begun to be recognized and documented. If you have HPP, tell your eye doctor to look for these four ocular findings of concern.

What to Keep an Eye out for

Ectopic Calcification of Conjunctiva

In non-medical language, this simply means calcium deposits on the conjunctiva. Which is the membrane surrounding the white of the eye and the underside of the eyelids. Sometimes this finding could be a result of treatment with asfotase alfa. **The deposits are benign, rarely cause irritation, and do not affect vision.**

Band Keratopathy

Like ectopic calcification of conjunctiva band keratopathy also involves calcium deposits, but in this case the deposits are on the cornea, presenting a more complicated and potentially more serious outcome. If severe, **it can interfere with vision and may require chemical removal.**

Optic Atrophy

A finding of optic atrophy is **an indication of damage to the optic nerve, which can lead to irreparable vision loss.** Children with HPP are at especially high risk for this outcome, as optic atrophy can be a downstream result of HPP-related craniosynostosis, premature fusion of the skull.

Angioid Streaks

A finding of angioid streaks indicates an abnormal calcification and brittling of the delicate membrane between the retina and choroid (the vascular layer of the eye). The risk is that new, abnormal blood vessels can form and rupture, **leading to significant loss of vision.**



How To Protect Your Eye Health

- **Get an annual eye examination with an optometrist (O.D.) or ophthalmologist (M.D.) every one to two years.** Dilation must be part of the exam. Your eye doctor is likely to have limited knowledge of HPP in general and its relationship to certain eye problems, but will have the knowledge and skills to detect ocular issues. You might want to take this fact sheet to your eye doctor and suggest that he or she contact Soft Bones for more information.
- **Don't ignore sudden changes in vision.** Call your eye doctor as soon as possible.