OASIS DERMATOLOGY GROUP

Ph: (956) 971 - 0404
www.oasisderm.com

Pseudoxanthoma Elasticum

Pseudoxanthoma elasticum (PXE) is a rare, inherited connective tissue disorder affecting the skin, eyes, gastrointestinal (GI), and cardiovascular systems. Although the condition is typically present from childhood, clinical manifestations often do not appear until adolescence. Early recognition of PXE is crucial to managing and minimizing the long-term complications that can affect the cardiovascular and gastrointestinal systems.

Clinical Presentation

PXE usually presents with distinctive skin lesions that first appear on the neck, although lesions can also be found in other areas, such as the axillae, elbows, and groin. These lesions are typically asymptomatic but may be a cosmetic concern for patients. The characteristic skin lesions are small, yellowish papules, typically ranging from 1-5 mm in diameter, that develop in a linear or cobblestone pattern. As the disease progresses, these papules become more prominent, causing the skin to take on a "plucked chicken" or "gooseflesh" appearance.

In later stages, patients may experience other systemic manifestations due to the involvement of the cardiovascular and gastrointestinal systems. These include gastrointestinal bleeding, hypertension, chest pain, and hematuria. The severity of these symptoms can vary depending on the extent of organ involvement, and complications such as cardiovascular disease and GI bleeding can significantly impact the patient's quality of life.

Diagnosis

The diagnosis of PXE is primarily clinical, based on the characteristic skin lesions and the patient's medical history. However, several diagnostic tests can be performed to confirm the diagnosis and assess for systemic involvement:

- ***Skin Biopsy****:* Histopathological examination of a skin biopsy is essential for diagnosis, revealing calcification and fragmentation of elastic fibers in the dermis, which is a hallmark of PXE.
- ***Genetic Testing****:* Molecular genetic testing for mutations in the *ABCC6* gene can provide definitive confirmation of the diagnosis.
- ***Blood Tests****:* Routine blood tests, including a complete blood count and metabolic panel, are often used to monitor systemic involvement, particularly renal function, which may be affected in PXE.

- ***Urinalysis and Stool Screening****:* These tests can help detect early signs of kidney and gastrointestinal involvement, such as hematuria or gastrointestinal bleeding.
- ***Ophthalmologic Evaluation****:* Given the potential for retinal and choroidal involvement, regular eye exams are recommended to monitor for changes in vision and detect early ocular complications.

Management

Currently, there is no cure for PXE, and much of the management focuses on addressing the symptoms and preventing or minimizing complications. Treatment strategies for PXE can be grouped into conservative management, surgical interventions, and regular monitoring for systemic involvement.

- ***Symptomatic Management****:*
  - ***Skin Lesions****:* While skin changes are irreversible, cosmetic concerns can be addressed through surgical removal of redundant skin folds, especially in cases where skin sagging occurs later in the disease course. Collagen and autologous fat injections may be used for treatment of facial wrinkles or creases, particularly in the forehead region.
  - ***Cardiovascular and Gastrointestinal Care****:* Regular consultations with a cardiologist are recommended for monitoring cardiovascular health, as patients with PXE are at increased risk of hypertension, coronary artery disease, and peripheral vascular disease. Gastrointestinal monitoring, particularly for gastrointestinal bleeding, is essential, as some patients may experience complications such as bleeding ulcers or esophageal rupture due to abnormal mineralization of the gastrointestinal tissues.
- ***Ophthalmologic Surveillance****:*
  - Routine visits to an ophthalmologist are critical to detect ocular complications, such as angioid streaks in the retina, which can lead to vision loss if not managed appropriately. Early detection allows for interventions that may preserve vision, including laser treatment for choroidal neovascularization if present.
- ***Pharmacologic Treatment****:*
  - Although no specific pharmacologic therapies are approved for PXE, some evidence suggests that bisphosphonates, which are commonly used to treat osteoporosis, may help reduce the deposition of calcium in tissues and prevent the progression of skin and cardiovascular complications. Further research is needed to establish the efficacy and safety of bisphosphonates in PXE.
- ***Preventive Care****:*
  - Preventing complications such as cardiovascular events or gastrointestinal bleeding involves the use of standard treatments for hypertension and vascular disease. Early intervention for any signs of renal or gastrointestinal involvement is also crucial to minimize further damage.

Prognosis

The prognosis of PXE depends largely on the extent of systemic involvement. While the skin lesions are typically harmless, the cardiovascular and gastrointestinal manifestations can lead to significant morbidity. Ocular involvement, particularly the development of angioid streaks, may result in vision loss, but with appropriate monitoring and intervention, these outcomes can be mitigated. Overall, with early recognition and management, many patients with PXE can live a normal life expectancy, although close monitoring of cardiovascular, gastrointestinal, and ophthalmologic health is essential.

Conclusion

Pseudoxanthoma elasticum (PXE) is a rare genetic disorder that causes progressive degeneration of elastic fibers in various tissues, leading to characteristic skin lesions and potential complications in the eyes, cardiovascular system, and gastrointestinal tract. Diagnosis is based on clinical findings, genetic testing, and biopsy, while management focuses on symptomatic treatment and preventive care. Although there is no cure for PXE, regular monitoring and interventions can significantly improve quality of life and prevent severe complications. Future research may provide new insights into potential therapies for PXE, with ongoing efforts to better understand the underlying pathophysiology and improve patient care.

References

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