# Incontinentia Pigmenti

- Pott's puffy tumor with intracranial extension in a child with incontinentia pigmenti: case based review of the eponymous disease
- Incontinentia pigmenti with intracranial arachnoid cyst: A case report
- Introduction to phacomatoses (neurocutaneous disorders) in childhood
- Phacomatosis and genetically determined tumors: the transition from childhood to adulthood

Incontinentia Pigmenti (IP) is a rare genetic disorder that primarily affects the skin, hair, teeth, nails, and central nervous system. It is also known as Bloch-Sulzberger syndrome. The condition is named for the characteristic patterns of skin changes that occur in affected individuals, particularly the swirling patterns of hyperpigmentation seen in the skin.

Causes Incontinentia pigmenti is caused by mutations in the IKBKG gene (also known as NEMO gene), which is located on the X chromosome. The IKBKG gene provides instructions for making a protein that plays a key role in protecting cells from undergoing apoptosis (cell death) in response to various signals. This protein is also involved in the regulation of inflammation and the immune response.

X-linked Dominant Pattern: IP follows an X-linked dominant inheritance pattern. It predominantly affects females, as males with the condition usually do not survive to birth due to the importance of the IKBKG gene in early development. In rare cases, males with a milder form of the mutation can survive, often due to genetic mosaicism (where some cells in the body have the mutation and others do not). Symptoms and Stages The symptoms of Incontinentia Pigmenti are diverse and can vary greatly among individuals, even within the same family. The disorder usually affects the skin, and its symptoms tend to follow four distinct stages:

Stage 1 - Blistering (Vesicular Stage):

This stage typically occurs in newborns or within the first few weeks of life. It is characterized by the appearance of blister-like lesions on the skin, which often form in linear or swirling patterns. These blisters may be present on the arms, legs, and trunk and are typically filled with clear or yellowish fluid. The blisters usually heal over weeks to months and leave behind red or dark brown streaks or patches. Stage 2 - Wart-like (Verrucous Stage):

During this stage, which typically occurs in early infancy, the skin lesions evolve into rough, wart-like growths. These lesions are often found on the limbs and may persist for several months before resolving. Stage 3 - Hyperpigmentation (Hyperpigmented Stage):

This stage generally begins in infancy or early childhood and can last for several years. It is characterized by the development of swirled or streaked areas of dark pigmentation, usually following the lines of Blaschko (patterns of skin cells migration during embryonic development). These pigmented areas are commonly seen on the trunk and the extremities. Stage 4 - Hypopigmentation (Atrophic Stage):

In later childhood or adulthood, affected skin areas may become pale or atrophic (thinned). The pigmentation may fade, leaving areas of lighter skin (hypopigmentation) or areas with diminished hair growth. Other Symptoms Apart from skin manifestations, IP can affect other parts of the body:

Hair: Individuals with IP may have abnormalities of the hair, including sparse scalp hair, alopecia (hair loss), or brittle hair. Teeth: Dental anomalies are common, including missing teeth (hypodontia), small teeth (microdontia), or abnormal spacing. Nails: Nail changes can include ridges, pitting, or

abnormally shaped nails. Eyes: Some individuals with IP may have eye abnormalities, such as strabismus (misaligned eyes), cataracts, retinal detachment, or other retinal vascular changes that can lead to vision impairment or blindness. Central Nervous System: Neurological complications can occur in about 30% of cases and may include seizures, developmental delay, intellectual disability, motor difficulties, or other neurological abnormalities. Diagnosis Diagnosis of Incontinentia Pigmenti is typically based on clinical findings, especially the characteristic skin changes. A skin biopsy can sometimes be used to confirm the diagnosis by showing specific histopathological features. Genetic testing can identify mutations in the IKBKG gene, confirming the diagnosis in most cases.

Management and Treatment There is no cure for Incontinentia Pigmenti, and treatment is primarily supportive and symptomatic:

Dermatological Care: Management of skin lesions, especially in the blistering stage, includes careful wound care to prevent infection and scarring. Ophthalmologic Monitoring: Regular eye exams are crucial to detect and manage any eye abnormalities early, especially those that might threaten vision. Dental Care: Early dental evaluation and orthodontic interventions may be necessary for those with dental anomalies. Neurological Support: Children with neurological symptoms may benefit from physical therapy, occupational therapy, and other supportive measures. Prognosis The prognosis for individuals with Incontinentia Pigmenti is variable and depends largely on the severity of the symptoms and the presence of complications. While many individuals have a normal life expectancy and lead healthy lives, those with severe neurological or ocular involvement may face significant challenges. Regular follow-up with a multidisciplinary team is essential to manage the condition effectively.

## Case reports

A case of Pott's puffy tumor presenting in a child with IP. We also performed a literature review of reported cases of PPT associated with immune dysfunction. We discuss the clinical presentation, diagnosis, and management of these lesions.

Results: We identified 12 cases of PPT associated with immune dysfunction/suppression. Diabetes was the most commonly identified cause followed by iatrogenic immunosuppression. Surgery is the standard treatment for managing PPT and the management of PPT with and without intracranial involvement, particularly in the context of underlying immune dysfunction/suppression, is discussed.

Conclusion: PPT remains a rare but not infrequent diagnosis, often requiring neurosurgical intervention. Immune dysfunction/suppression is an additional risk factor that may predispose to PPT. Early and aggressive management should be instituted for optimal outcome [1)]

1)

Datta D, Tu A. Pott's puffy tumor with intracranial extension in a child with incontinentia pigmenti: case based review of the eponymous disease. Childs Nerv Syst. 2024 Aug 24. doi: 10.1007/s00381-024-06577-4. Epub ahead of print. PMID: 39180698.