Peters Anomaly

An abnormality of embryologic cell line cell migration
A

- Peters Anomaly
  - An abnormality of neural-crest cell migration
Q

• Peters Anomaly
  • An abnormality of neural-crest cell migration

*What are neural crest cells?*
Q/A

Peters Anomaly

An abnormality of neural-crest cell migration

*What are neural crest cells?*

A special subpopulation of embryologic cell type cells that migrate across the embryo and deposit themselves at a wide variety of locations, eventually differentiating into many distinct tissues.
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What is the term for a condition arising from the abnormal migration or differentiation of neural-crest cells?
Peters Anomaly

- An abnormality of neural-crest cell migration

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**What is the term for a condition arising from the abnormal migration or differentiation of neural-crest cells?**
A neurocristopathy
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Neural-crest-cell migration concerning the anterior segment occurs in three ‘waves.’
Which wave involves which future structure?
First wave →
Second wave →
Third wave →
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Second wave ➔
Third wave ➔
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First wave ➔ Corneal endothelium
Second wave ➔ Iris stroma — epithelium?
Third wave ➔ Corneal stroma (keratocytes)

What about the iris epithelium—is it neural-crest derivative as well?
No, it derives from the neuroectodermal cells that line the optic vesicle. In addition to the iris epithelium, the optic vesicle gives rise to the ciliary body epithelium, as well as to the optic nerve, RPE, and retina.

How many epithelial layers are present on the posterior iris?
Two

By definition, epithelial cells rest on a basement membrane, and thus have an orientation—specifically, a basal portion (= the side in contact with the BM) and an apex (the side opposite). With respect to one another, how are the two iris epithelial layers oriented?
Apex to apex

Of the two iris epithelial layers, which is/are pigmented?
Both are pigmented.
Q/A

- Peters Anomaly

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**What about the iris epithelium—is it neural-crest derivative as well?**
No, it derives from the cells that line the (the embryologic outpouching off of the) two words

**Two words**

**Two words**
A

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When do neural-crest cells swoop in to form the primordial corneal epithelium?

First wave → Corneal endothelium
Second wave → Iris stroma
Third wave → Corneal stroma (keratocytes)
A later wave → Corneal epithelium?
Q/A

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  - An abnormality of neural-crest cell migration

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They don’t. The corneal epithelium derives from the surface ectoderm of the outer ‘body’ wall of the embryo.

Third wave ➔ Corneal stroma (keratocytes)
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Vexed by all this embryology? There’s a slide-set for that (FELT21)

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- An abnormality of neural-crest cell migration
- A form of anterior segment dysgenesis
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Let’s take a moment to overview the anterior segment dysgeneses. We divvy them up into two broad categories based on a very basic anatomic consideration—what is that consideration?
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To which category does Peters anomaly belong?
Let’s take a moment to overview the anterior segment dysgeneses. We divvy them up into two broad categories based on a very basic anatomic consideration—what is that consideration? It is whether the dysgenesis involves the **peripheral** vs **central** portion of the anterior segment.

To which category does Peters anomaly belong? Peters is a **central** form of anterior segment dysgenesis.
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To which category does Peters anomaly belong?
Peters is a central form of anterior segment dysgenesis.

What is the other classic form of central anterior segment dysgenesis?
Let’s take a moment to overview the anterior segment dysgeneses. We divvy them up into two broad categories based on a very basic anatomic consideration—what is that consideration? It is whether the dysgenesis involves the peripheral vs central portion of the anterior segment.

To which category does Peters anomaly belong? Peters is a central form of anterior segment dysgenesis.

What is the other classic form of central anterior segment dysgenesis? Posterior keratoconus (Note: This is nothing like the anterior form you’re familiar with)
Let’s take a moment to overview the anterior segment dysgeneses. We divvy them up into two broad categories based on a very basic anatomic consideration—what is that consideration? It is whether the dysgenesis involves the peripheral vs central portion of the anterior segment.

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**Posterior keratoconus** (Note: This is nothing like the anterior form you’re familiar with)

What are the two classic forms of peripheral anterior segment dysgenesis?
**Posterior embryotoxon** and **Axenfeld-Rieger syndrome**
Let's take a moment to overview the anterior segment dysgeneses. We divvy them up into two broad categories based on a very basic anatomic consideration—what is that consideration? It is whether the dysgenesis involves the **peripheral** vs **central** portion of the anterior segment.

To which category does Peters anomaly belong?
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What is the other classic form of **central** anterior segment dysgenesis?
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What are the two classic forms of **peripheral** anterior segment dysgenesis?
**Posterior embryotoxon** and **Axenfeld-Rieger syndrome**
Peters Anomaly

- An abnormality of neural-crest cell migration
- A form of anterior segment dysgenesis
- Hallmarks:
  1) A central corneal
t
A

- **Peters Anomaly**
  - An abnormality of neural-crest cell migration
  - A form of *anterior segment dysgenesis*
  - Hallmarks:
    1) A central corneal opacity

Peters Anomaly

- An abnormality of neural-crest cell migration
- A form of anterior segment dysgenesis
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What's the classic Peters presentation?
Peters Anomaly

- An abnormality of neural-crest cell migration
- A form of anterior segment dysgenesis
- Hallmarks:
  1) A central corneal opacity

What's the classic Peters presentation? As a cloudy cornea at birth (it's in the STUMPED mnemonic)
Q

- **Peters Anomaly**
  - An abnormality of neural-crest cell migration
  - A form of anterior segment dysgenesis
  - Hallmarks:
    1) A **central corneal opacity**

**What’s the classic Peters presentation?**
As a cloudy cornea at birth (it’s in the STUMPED mnemonic)

**Note:** There are two S’s and two E’s

**What is the STUMPED mnemonic** for a cloudy cornea in an infant?

- S: **S**clerocornea
- T: **T**rauma (e.g., forcep injury)
- U: **U**lcer
- M: **M**ucopolysaccharidosis
- P: **P**eters anomaly
- E: **E**levated IOP (congenital glaucoma)
- D: **D**ermoid of the cornea
Peters Anomaly

- An abnormality of neural-crest cell migration
- A form of anterior segment dysgenesis

Hallmarks:

1) A central corneal opacity

What is the STUMPED mnemonic for a cloudy cornea in an infant?
- Sclerocornea; Stromal dystrophy (CHSD)
- Trauma (eg, forcep injury)
- Ulcer
- Mucopolysaccharidosis
- Peters anomaly
- Endothelial dystrophy (CHED); Elevated IOP (congenital glaucoma)
- Dermoid of the cornea

Note: There are two S’s and two E’s
Peters Anomaly

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- Ulcer
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- Endothelial dystrophy (CHED); Elevated IOP (congenital glaucoma)
- Dermoid of the cornea

In the present context, what do these acronyms stand for?
- CHSD:
- CHED:
Peters Anomaly

- An abnormality of neural-crest cell migration
- A form of *anterior segment dysgenesis*
- Hallmarks:
  1) A **central corneal opacity**

*What’s the classic Peters presentation?* As a cloudy cornea at birth (it’s in the **STUMPED mnemonic**)

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What's the classic Peters presentation? As a cloudy cornea at birth (it's in the STUMPED mnemonic)

How cloudy is cloudy?
A

- Peters Anomaly
  - An abnormality of neural-crest cell migration
  - A form of anterior segment dysgenesis
  - Hallmarks:
    1) A central corneal opacity

What's the classic Peters presentation? As a cloudy cornea at birth (it's in the STUMPED mnemonic)

How cloudy is cloudy? The opacity ranges in severity from a faint haze to an opaque, elevated and vascularized mess
Peters anomaly: Cloudy cornea
Q

- Peters Anomaly
  - An abnormality of neural-crest cell migration
  - A form of anterior segment dysgenesis
  - Hallmarks:
    1) A central corneal opacity
    2) Iris-cornea and/or lens-cornea
Peters Anomaly

- An abnormality of neural-crest cell migration
- A form of anterior segment dysgenesis
- Hallmarks:
  1. A central corneal opacity
  2. Iris-cornea and/or lens-cornea adhesions
1. Defect of the posterior central cornea, including the absence of Descemet’s and subjacent endothelium

2. Adhesions extending from the iris to the posterior corneal defect

Peters anomaly: Iris-cornea adhesions
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What is the term for a condition arising from the abnormal migration or differentiation of neural-crest cells?

A neurocristopathy

Neural-crest-cell migration concerning the anterior segment occurs in three ‘waves.’ Which wave involves which future structure?

First wave \(\rightarrow\) Corneal endothelium
Second wave \(\rightarrow\) Iris stroma
Third wave \(\rightarrow\) Corneal stroma (keratocytes)

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  1) A central corneal opacity
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Recall the question below…Now you can see how the main hallmarks of Peters anomaly derive from a failure of first-wave migration to properly cleave between the primitive cornea and lens.

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There are two subtypes of Peters—what are they called?

Central

Posterior

Keratoconus

Peters anomaly

Two subtypes
Peters Anomaly

- An abnormality of neural-crest cell migration
- A form of anterior segment dysgenesis
- Hallmarks:
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There are two subtypes of Peters—what are they called? They are called ‘Type 1 and Type 2’
Peters Anomaly

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1) A central corneal opacity
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There are two subtypes of Peters—what are they called? They are called ‘Type 1 and Type 2’

How do they differ clinically?
Type 1:
Type 2:

Central

Peters anomaly

Type 1
Type 2
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There are two subtypes of Peters—what are they called? They are called ‘Type 1 and Type 2’

How do they differ clinically?
Type 1: Iridocorneal adhesions present
Type 2: Corneolenticular adhesions present

Central

Peters anomaly

Type 1
Type 2
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Type 2:
Peters Anomaly

- An abnormality of neural-crest cell migration
- A form of anterior segment dysgenesis
- Hallmarks:
  1) A central corneal opacity
  2) Iris-cornea and/or lens-cornea adhesions

There are two subtypes of Peters—what are they called? They are called ‘Type 1 and Type 2’

How do they differ clinically?
Type 1: Iridocorneal adhesions present
Type 2: Corneolenticular adhesions present
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Which type carries a more ominous ophthalmic prognosis?

Type 1
Type 2
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Which type carries a more ominous ophthalmic prognosis? Type 2
Peters anomaly Type 2: Lens-cornea adhesions
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Hallmarks:
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3) Anterior
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  3) Anterior *cataract*

Which specific types of anterior cataract?
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Which specific types of anterior cataract?
Anterior cortical, and anterior polar
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  4) Misshapen lens
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• **Peters Anomaly**
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If the ‘misshapen’ lens were smaller and rounder than normal, what particular condition would that evoke?

Microspherophakia

Is microspherophakia associated with Peters anomaly?
Yes (although only “occasionally” per the BCSC Lens book)
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- **Microspherophakia**

*Is microspherophakia associated with Peters anomaly?*
Yes (although only “occasionally” per the BCSC Lens book)
Peters anomaly: Small, cataractous and misshapen lens
This image shows complete corneal opacification thought clinically to be sclerocornea

Peters anomaly
This image shows complete corneal opacification thought clinically to be sclerocornea.

UBM of the same eye shows keratolenticular adhesion (ILA), aniridia with an iris stump (IR), and a small lens, revealing the correct diagnosis as Peters anomaly. This case demonstrates the importance of a complete anterior segment exam with UBM to make an accurate diagnosis.

Peters anomaly
Peters Anomaly

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A form of anterior segment dysgenesis

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Usually unilateral, usually sporadic
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- Usually *bilateral*, usually *sporadic*
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Bilateral cases → do this, doc
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- Peters-plus syndrome: Peters anomaly + short, short, abnormal including and abnormalities
Peters Anomaly

An abnormality of **neural-crest** cell migration

A form of **anterior segment dysgenesis**

Hallmarks:
1) A central corneal **opacity**
2) Iris-cornea and/or lens-cornea **adhesions**
3) Anterior **cataract**
4) **Misshapen** lens

Usually **bilateral**, usually **sporadic**

Bilateral cases → **complete genetic workup**

**Peters-plus** syndrome: Peters anomaly + short **stature**, short **digits**, abnormal **facies** including **cleft lip/palate** and **external ear** abnormalities
Peters Plus syndrome: Cleft lip/palate

Peters plus syndrome
Facial features of four patients with Peters Plus syndrome. Note the Peters anomaly, the long face, and the Cupid’s bow shape of the upper lip in all patients. Patients B and D have a repaired cleft lip and/or palate. Patient A is female; the rest are male.
Peters Plus syndrome: Cleft lip/palate

Peters plus syndrome