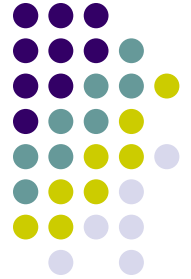


## Aicardi Syndrome



*What findings comprise the **Aicardi syndrome**?*

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Mnemonic is...

## Aicardi Syndrome



What findings comprise the **Aicardi syndrome**?

--A

--I

--C

--A

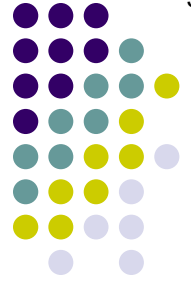
--R

--D

--I

Mnemonic is...*AICARDI*

## Aicardi Syndrome



What findings comprise the **Aicardi syndrome**?

--*Aicardi syndrome*

--*Includes...*

--**C**

--A

--R

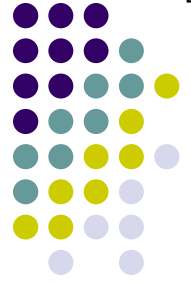
--D

--I



Mnemonic is...*AICARDI*

## Aicardi Syndrome



What findings comprise the **Aicardi syndrome**?

--*Aicardi syndrome*

--*Includes...*

--**Coloboma of**

abb.



--*A*

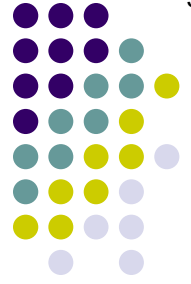
--*R*

--*D*

--*I*

Mnemonic is...*AICARDI*

## Aicardi Syndrome



What findings comprise the **Aicardi syndrome**?

--*Aicardi syndrome*

--*Includes...*

--**Coloboma of ONH**



--*A*

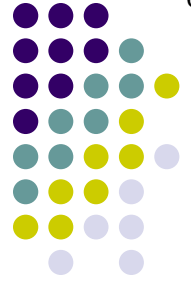
--*R*

--*D*

--*I*

Mnemonic is...*AICARDI*

## Aicardi Syndrome



What findings comprise the **Aicardi syndrome**?

--*Aicardi syndrome*

--*Includes...*

--**Coloboma of ONH**

--**A**

--*R*

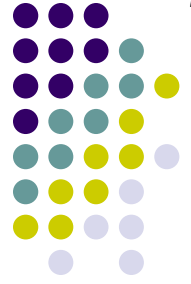
--*D*

--*I*



Mnemonic is...*AICARDI*

## Aicardi Syndrome



7

What findings comprise the **Aicardi syndrome**?

--*Aicardi syndrome*

--*Includes...*

--**Coloboma of ONH**

--**Absence of the**

two words

next

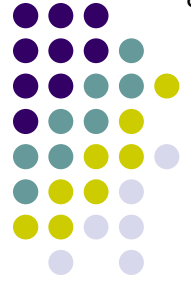
--*R*

--*D*

--*I*

Mnemonic is...*AICARDI*

## Aicardi Syndrome



What findings comprise the **Aicardi syndrome**?

--*Aicardi syndrome*

--*Includes...*

--**Coloboma of ONH**

--**Absence of the corpus callosum**



--*R*

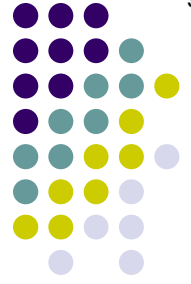
--*D*

--*I*

Mnemonic is...*AICARDI*



## Aicardi Syndrome



What findings comprise the **Aicardi syndrome**?

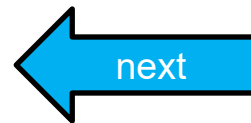
--*Aicardi syndrome*

--*Includes...*

--**Coloboma of ONH**

--**Absence of the corpus callosum**

--**R**



--**D**

--**I**

Mnemonic is...*AICARDI*

## Aicardi Syndrome



What findings comprise the **Aicardi syndrome**?

--*Aicardi syndrome*

--*Includes...*

--**Coloboma of ONH**

--**Absence of the corpus callosum**

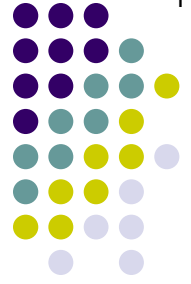
--**Retardation** ← next

--*D*

--*I*

Mnemonic is...*AICARDI*

## Aicardi Syndrome



What findings comprise the **Aicardi syndrome**?

--*Aicardi syndrome*

--*Includes...*

--**Coloboma of ONH**

--**Absence of the corpus callosum**

--**Retardation**

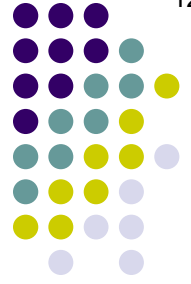
--**D**

--**/**



Mnemonic is...*AICARDI*

## Aicardi Syndrome



What findings comprise the **Aicardi syndrome**?

--*Aicardi syndrome*

--*Includes...*

--**Coloboma of ONH**

--**Absence of the corpus callosum**

--**Retardation**

--**Depigmented**

two words



--/

Mnemonic is...*AICARDI*

## Aicardi Syndrome



What findings comprise the **Aicardi syndrome**?

--*Aicardi syndrome*

--*Includes...*

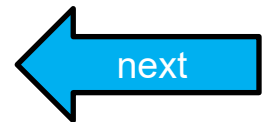
--**Coloboma of ONH**

--**Absence of the corpus callosum**

--**Retardation**

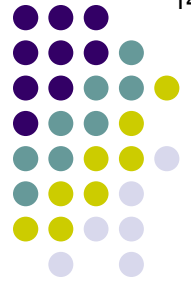
--**Depigmented chorioretinal lacunae**

--/



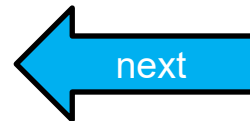
Mnemonic is...*AICARDI*

## Aicardi Syndrome



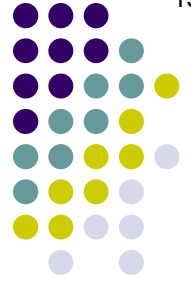
What findings comprise the **Aicardi syndrome**?

- Aicardi syndrome*
- Includes...*
- Coloboma of ONH**
- Absence of the corpus callosum**
- Retardation**
- Depigmented chorioretinal lacunae**
- /



Mnemonic is...*AICARDI*

## Aicardi Syndrome



What findings comprise the **Aicardi syndrome**?

--*Aicardi syndrome*

--*Includes...*

--**Coloboma of ONH**

--**Absence of the corpus callosum**

--**Retardation**

--**Depigmented chorioretinal lacunae**

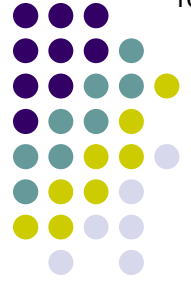
--**Infantile**

one word

next

Mnemonic is...*AICARDI*

## Aicardi Syndrome



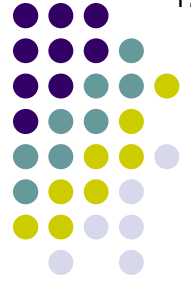
What findings comprise the **Aicardi syndrome**?

- Aicardi syndrome*
- Includes...*
- Coloboma of ONH**
- Absence of the corpus callosum**
- Retardation**
- Depigmented chorioretinal lacunae**
- Infantile seizures**



Mnemonic is...*AICARDI*





## *Aicardi Syndrome*

*What findings comprise the Aicardi syndrome?*

*--Aicardi syndrome*

*Next let's check out pics of the...*

*--Infantile seizures*

**--Coloboma of ONH**

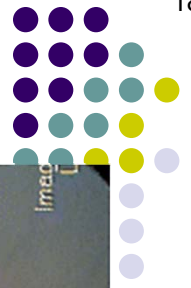
*--Absence of the corpus callosum*

*--Retardation*

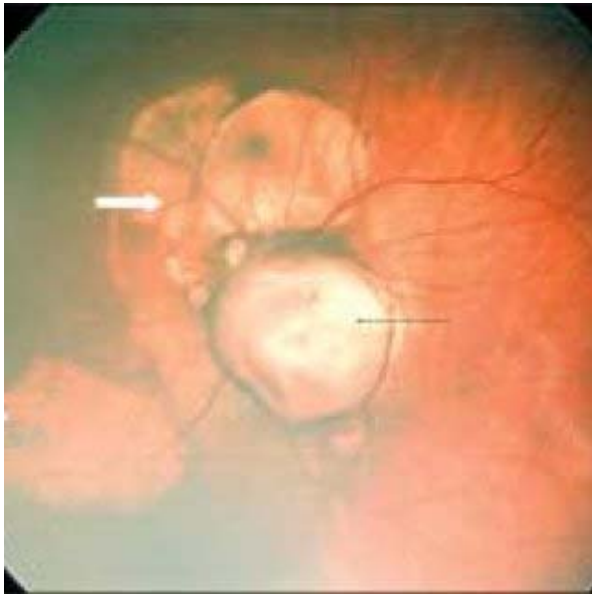
*--Depigmented chorioretinal lacunae*

*--Infantile seizures*

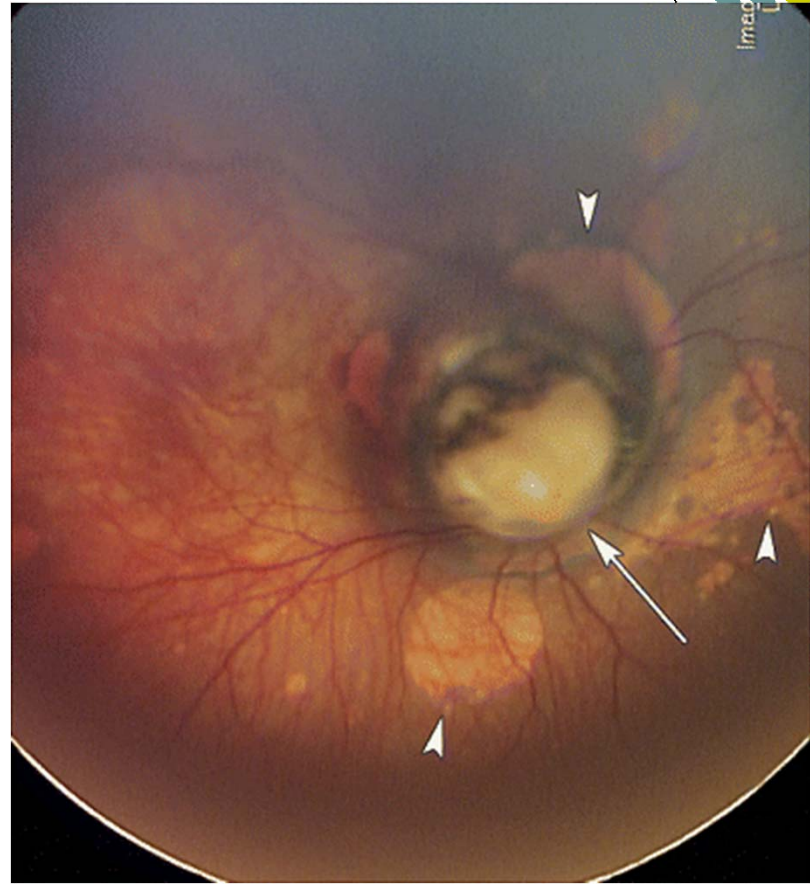
*Mnemonic is...AICARDI*



# Aicardi Syndrome



Note: The black arrow indicates the colobomatous ONH (the white arrow is pointing out a different finding we'll get to shortly)



Note: The white arrow indicates the colobomatous ONH (the white arrowheads are pointing out the same other finding as the white arrow in the other pic)

Aicardi syndrome: ONH coloboma

## Aicardi Syndrome



What findings comprise the Aicardi syndrome?

--Aicardi syndrome

Next let's check out pics of the...

--Includes...

--Coloboma of ONH

--**Absence of the corpus callosum**

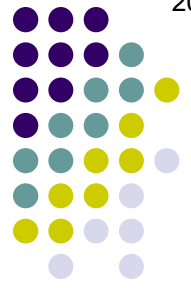
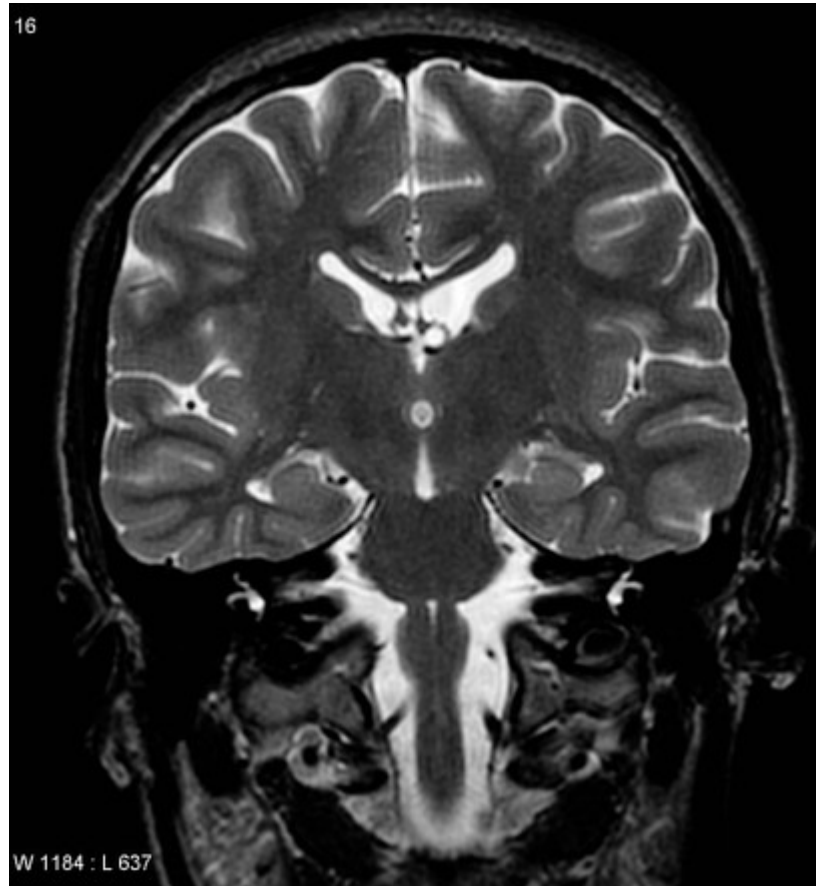
--Retardation

--Depigmented chorioretinal lacunae

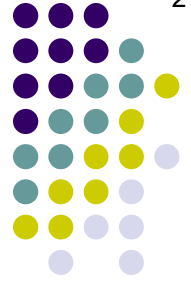
--Infantile seizures

Mnemonic is...AICARDI

## Aicardi Syndrome



Aicardi syndrome: Absence of the corpus callosum



## Aicardi Syndrome

What findings comprise the Aicardi syndrome?

--Aicardi syndrome

Next let's check out pics of the...

--Includes...

--Coloboma of ONH

--Absence of the corpus callosum

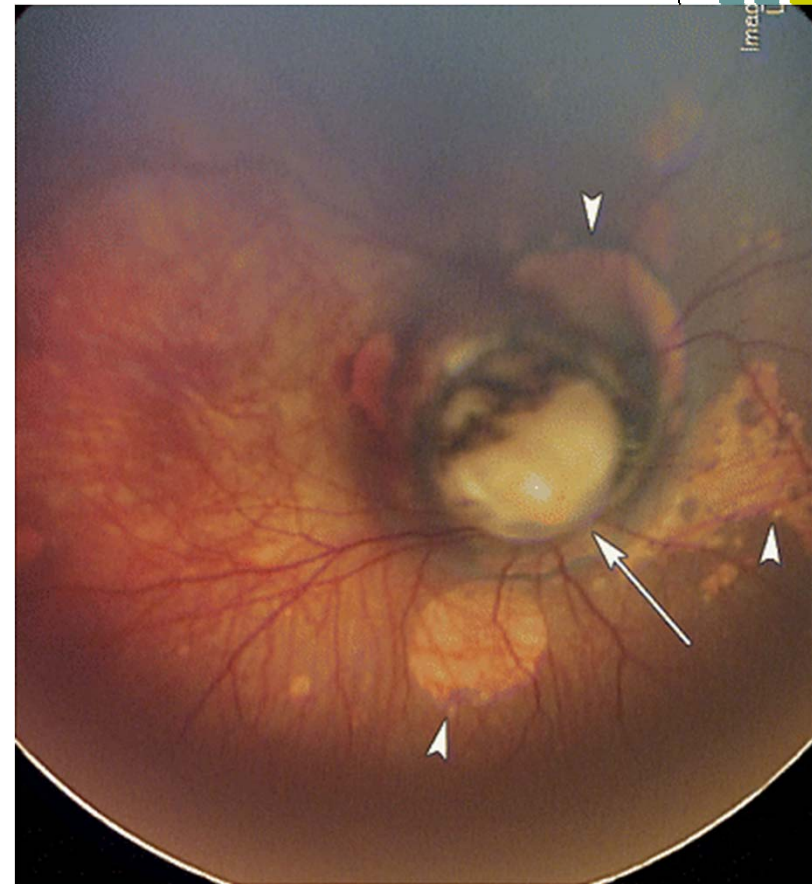
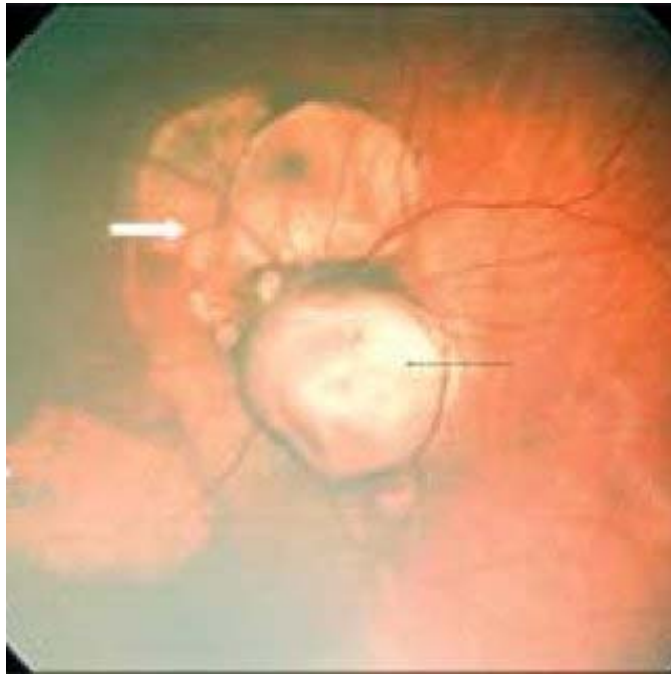
--Retardation

--**Depigmented chorioretinal lacunae**

--Infantile seizures

Mnemonic is...AICARDI

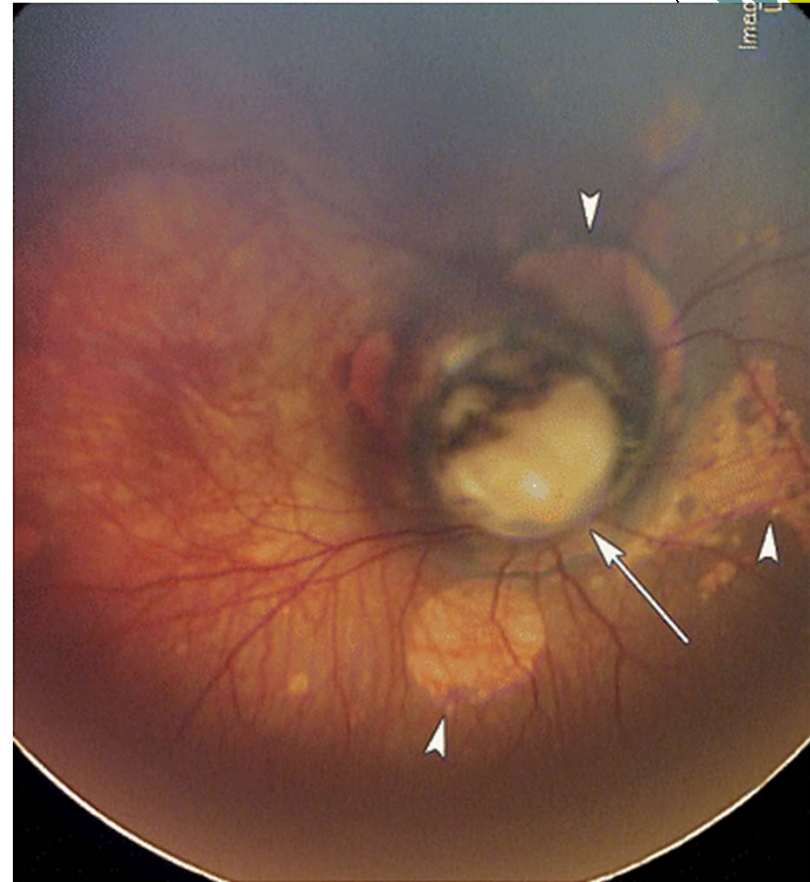
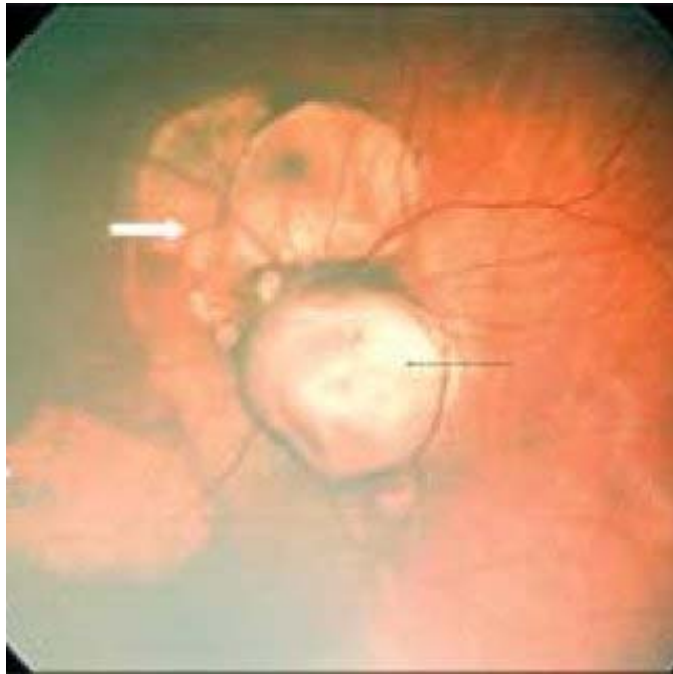
## Aicardi Syndrome



These pics were presented previously. It is to **depigmented chorioretinal lacunae** that the white arrow on the left and the white arrowheads on the right are pointing

Aicardi syndrome: Depigmented chorioretinal lacunae

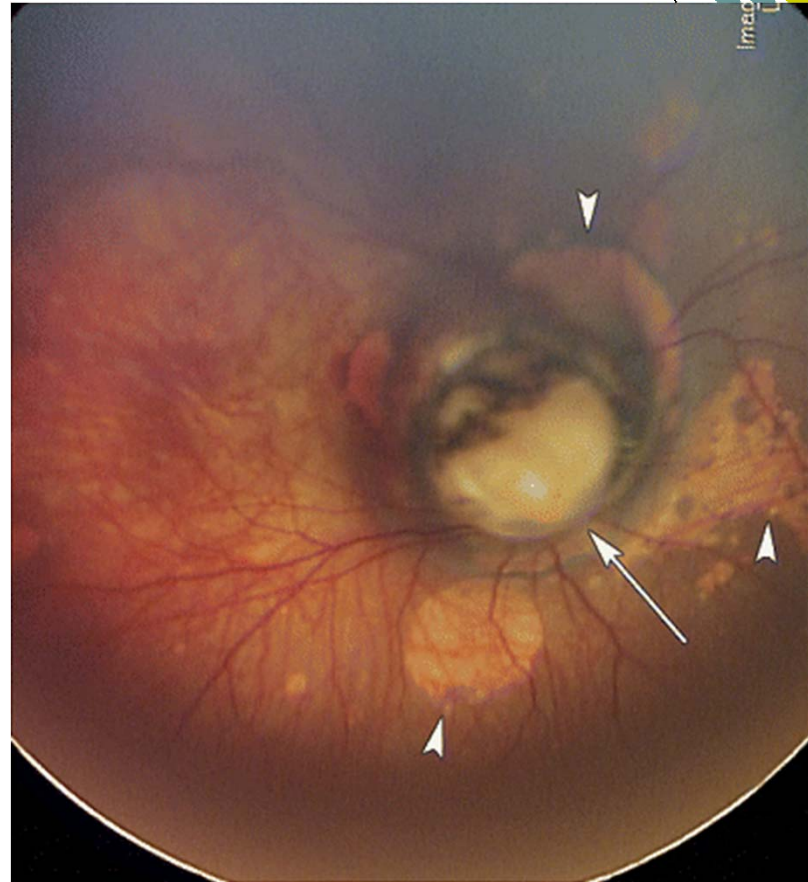
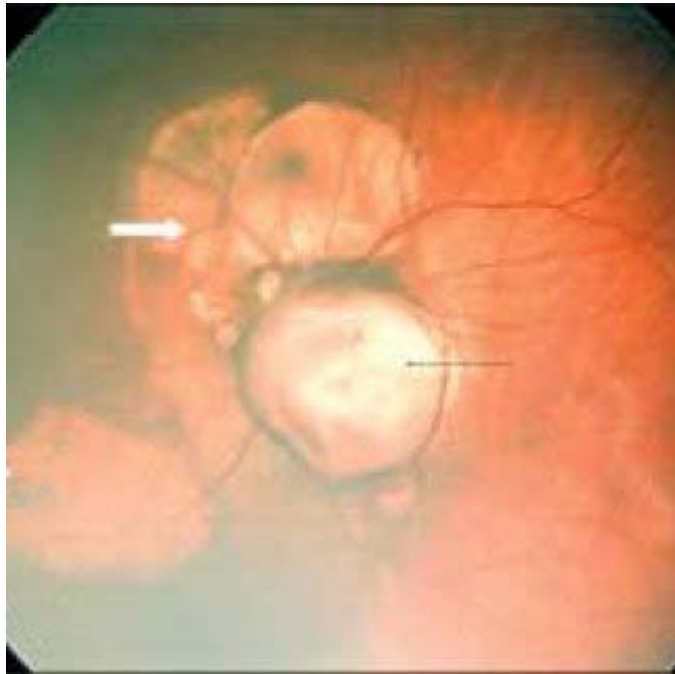
## Aicardi Syndrome



*At what location in the retina do the lacunae tend to be found?*

Aicardi syndrome. Depigmented chorioretinal lacunae

## Aicardi Syndrome

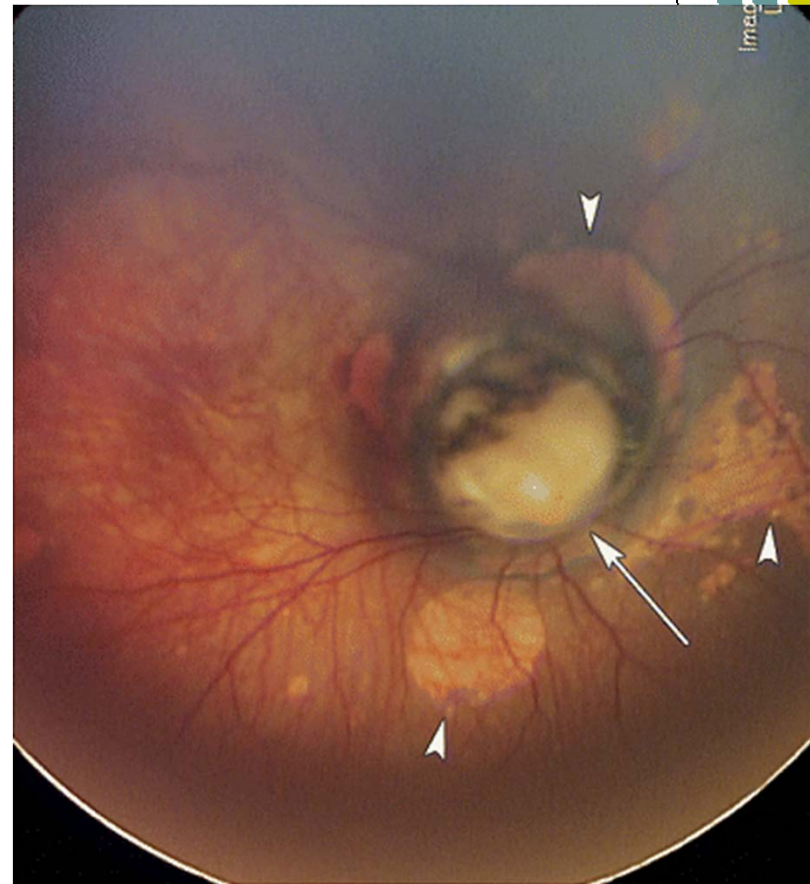
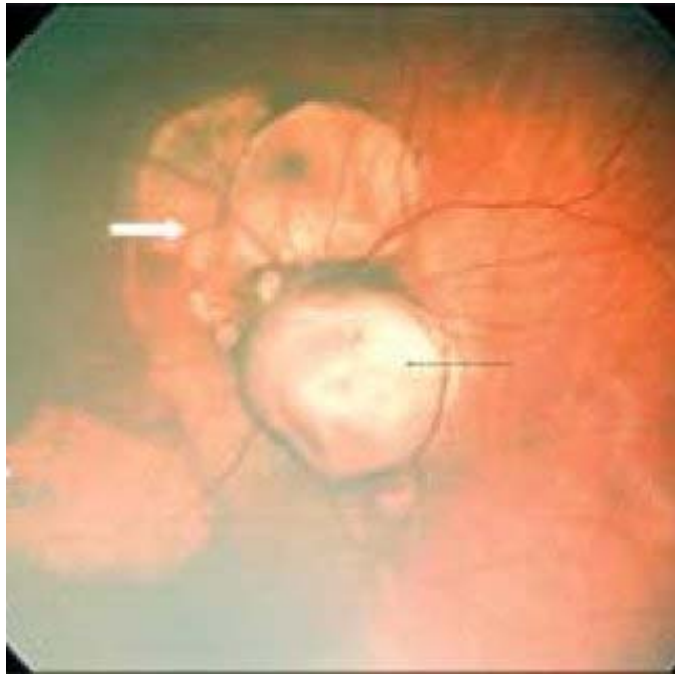


*At what location in the retina do the lacunae tend to be found?*  
The posterior pole, especially the   region

Aicardi syndrome. Depigmented chorioretinal lacunae



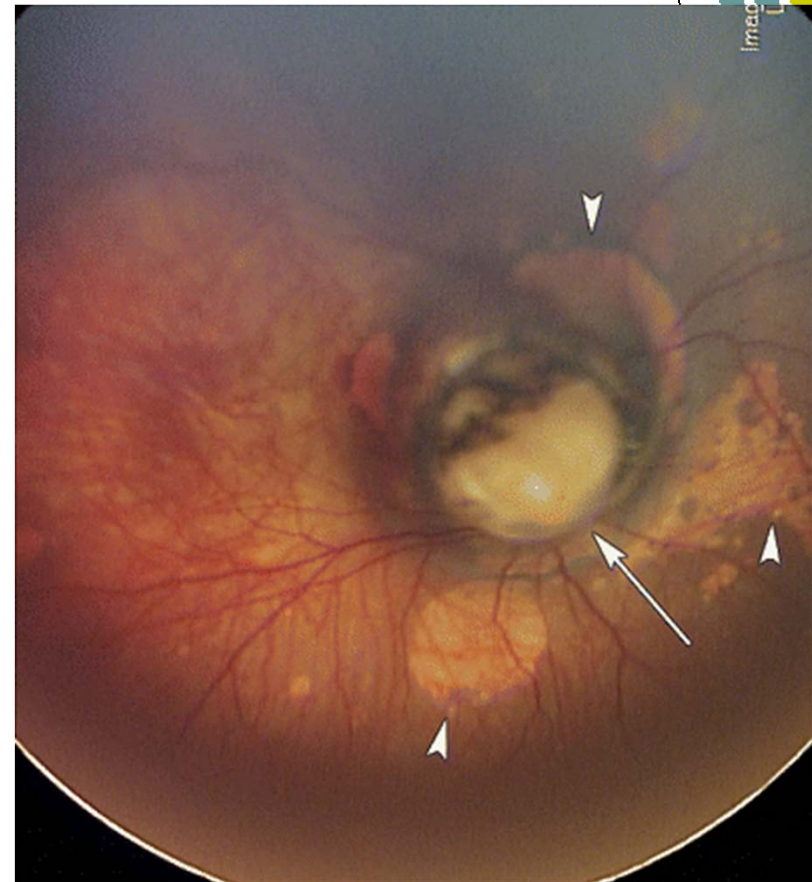
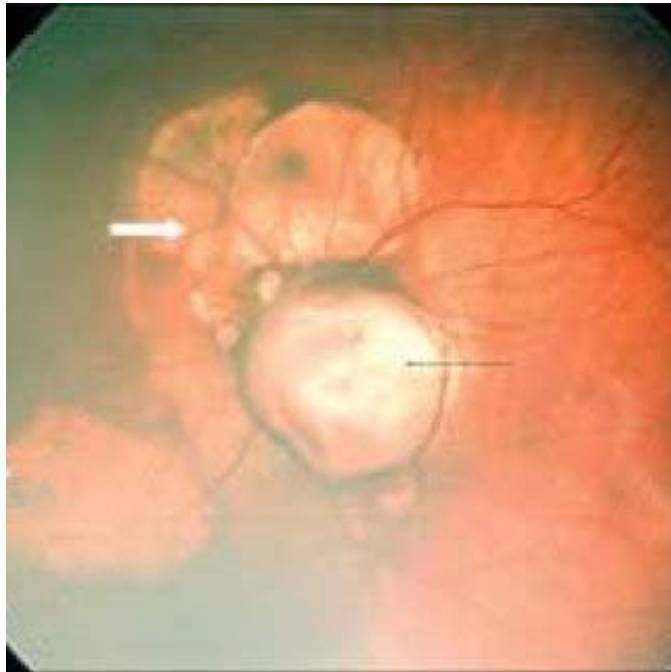
## Aicardi Syndrome



*At what location in the retina do the lacunae tend to be found?*  
The posterior pole, especially the peripapillary region

Aicardi syndrome. Depigmented chorioretinal lacunae

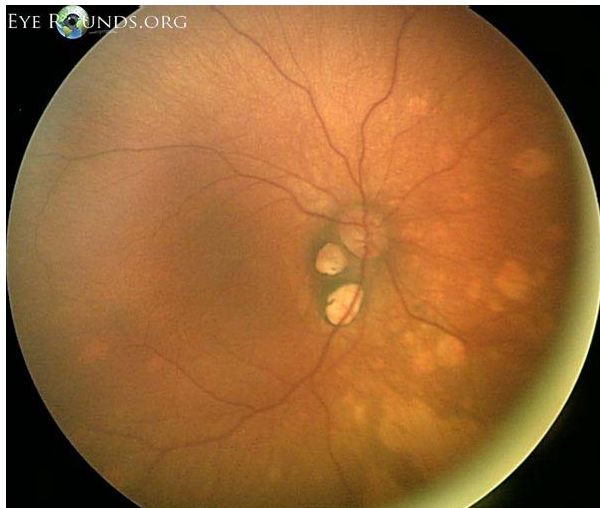
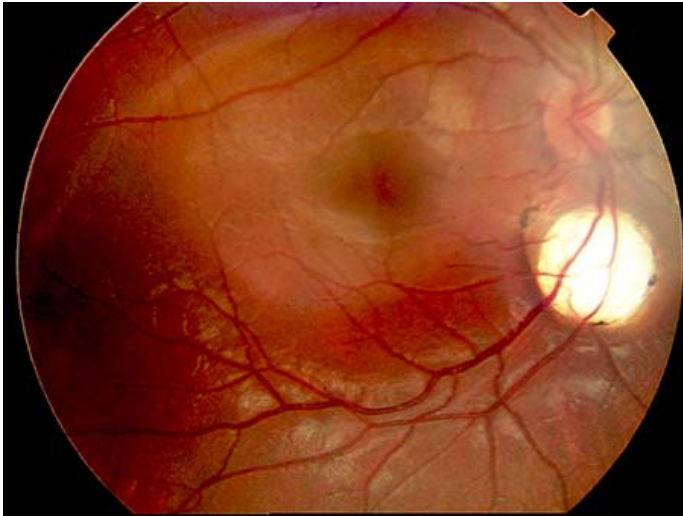
## Aicardi Syndrome



At what location in the retina do the lacunae tend to be found?  
 The posterior pole, especially the peripapillary region (it is not a coincidence that you can see the ONH in each of these pics)

Aicardi syndrome. Depigmented chorioretinal lacunae

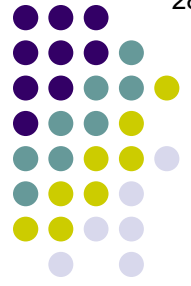
## Aicardi Syndrome



The depigmented chorioretinal lacunae are considered pathognomonic for Aicardi syndrome. The point being: If you commit only one pic to memory, make it of the depigmented lacunae!



## Aicardi Syndrome



What findings comprise the **Aicardi syndrome**?

--*Aicardi syndrome*

--*Includes...*

--**Coloboma of ONH**

--**Absence of the corpus callosum**

--**Retardation**

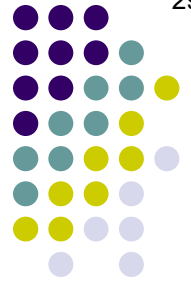
--**Depigmented chorioretinal lacunae**

--**Infantile seizures**

*Which three of these findings constitute the 'classic triad' of Aicardi syndrome?*

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--  
--

## Aicardi Syndrome



What findings comprise the **Aicardi syndrome**?

--Aicardi syndrome

--Includes...

--Coloboma of ONH

--**Absence of the corpus callosum**

--Retardation

--**Depigmented chorioretinal lacunae**

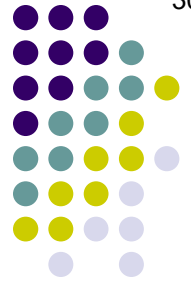
--**Infantile seizures**

Which three of these findings constitute the 'classic triad' of Aicardi syndrome?

--Absence of the corpus callosum

--Depigmented chorioretinal lacunae

--Infantile seizures



## Aicardi Syndrome

What findings comprise the **Aicardi syndrome**?

--Aicardi syndrome

--Includes...

--Coloboma of the eyes

--**Absence of the corpus callosum**

--Retardation

--Depigmented chorioretinal lacunae

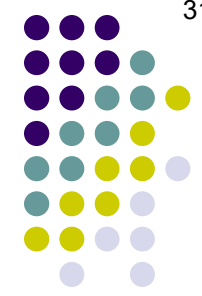
--Infantile seizures

Which three of these findings constitute the 'classic triad' of Aicardi syndrome?

--**Absence of the corpus callosum**

--Depigmented chorioretinal lacunae

Is the corpus callosum absence always complete?



# Aicardi Syndrome

What findings comprise the **Aicardi syndrome**?

--Aicardi syndrome

--Includes...

--Coloboma of the eyes

--**Absence of the corpus callosum**

--Retardation

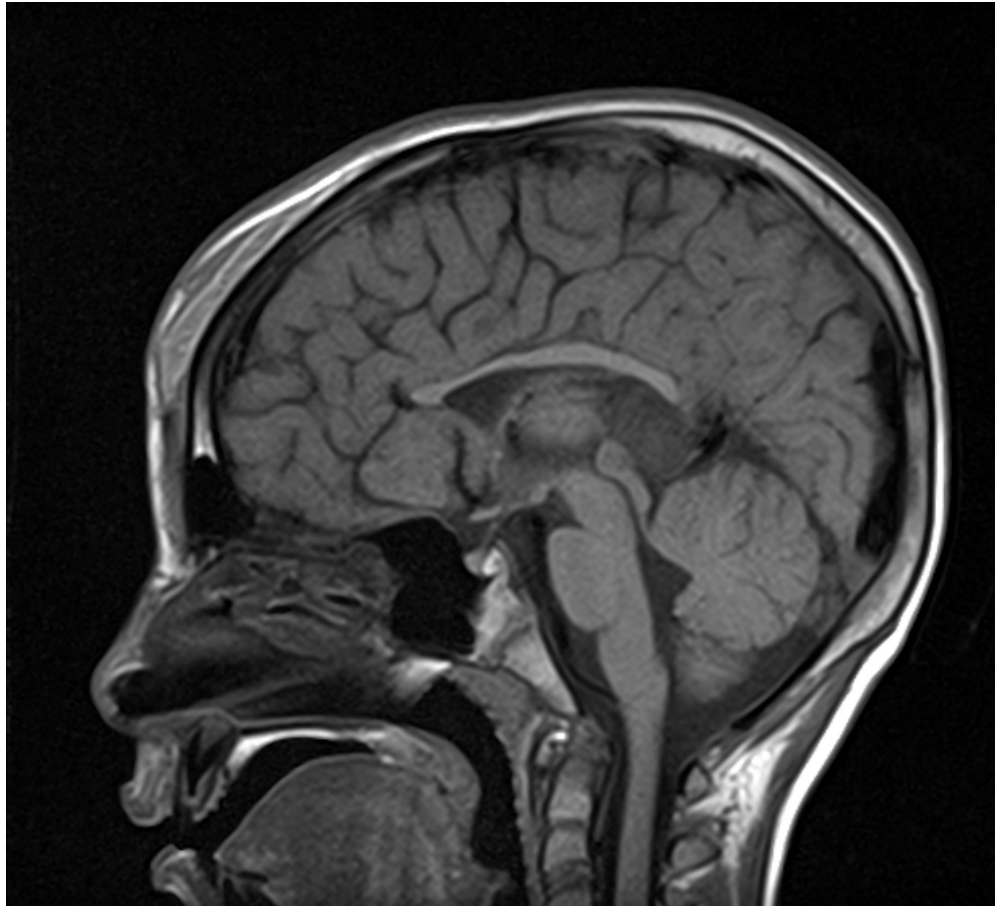
--Depigmented chorioretinal lacunae

--Infantile seizures

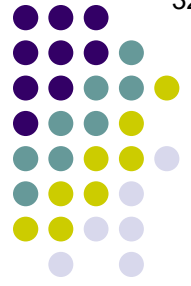
Which three of these findings constitute the 'classic triad' of Aicardi syndrome?  
--Absence of the corpus callosum  
--Depigmented chorioretinal lacunae

Is the corpus callosum absence always complete?  
No, in many cases it is partial

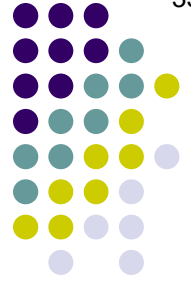
## *Aicardi Syndrome*



Partial agenesis of the corpus callosum in Aicardi syndrome





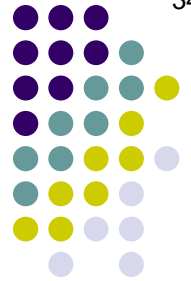


## **Aicardi Syndrome**

What findings comprise the **Aicardi syndrome**?

- *A*bsence of the corpus callosum
- *I*ntellectual disability
- *C*oloboma of ONH
- *A*bsence of the corpus callosum
- *R*etardation
- *D*epigmented chorioretinal lacunae
- *I*nfantile seizures

## Aicardi Syndrome



What findings comprise the **Aicardi syndrome**?

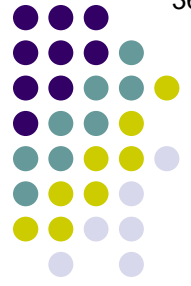
- *A* What is the inheritance pattern for Aicardi syndrome?  
X-linked dominant
- *I*
- *C* Coloboma of ONH
- *A* Absence of the corpus callosum
- *R* Retardation
- *D* Depigmented chorioretinal lacunae
- *I* Infantile seizures

## Aicardi Syndrome



What findings comprise the **Aicardi syndrome**?

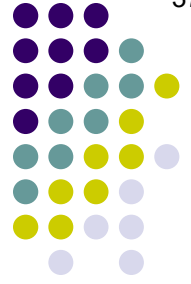
- *A* What is the inheritance pattern for Aicardi syndrome?  
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- *I* What is the gender predilection?
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- *A* Absence of the corpus callosum
- *R* Retardation
- *D* Depigmented chorioretinal lacunae
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## **Aicardi Syndrome**

What findings comprise the **Aicardi syndrome**?

- *A* What is the inheritance pattern for Aicardi syndrome?  
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- *I* What is the gender predilection?  
As it is lethal in males, the vast majority of pts are phenotypically female
- *C* Coloboma of ONH
- *A* Absence of the corpus callosum
- *R* Retardation
- *D* Depigmented chorioretinal lacunae
- *I* Infantile seizures



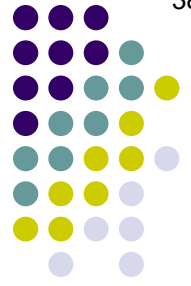
## Aicardi Syndrome

What findings comprise the **Aicardi syndrome**?

- *A* What is the inheritance pattern for Aicardi syndrome?  
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- *I* What is the gender predilection?  
As it is lethal in males, **the vast majority** of pts are phenotypically female
- *C* Coloboma of CNIII

'Vast majority'? If it's lethal in males, how do **any** survive to manifest the condition?

- *R* Retardation
- *D* Depigmented chorioretinal lacunae
- *I* Infantile seizures



## Aicardi Syndrome

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There are case reports of Aicardi syndrome in phenotypical males with 47,XXY karyotype

- *R* Retardation
- *D* Depigmented chorioretinal lacunae
- *I* Infantile seizures



# Aicardi Syndrome

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- *A* What is the inheritance pattern for Aicardi syndrome?  
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There are case reports of Aicardi syndrome in phenotypical males with **47,XXY karyotype**

- *R* Retardation
- *D* Depigmentation  
**47,XXY males are syndromic. What is the eponymous name for this syndrome?**
- *I* Infantile seizures



# Aicardi Syndrome

What findings comprise the **Aicardi syndrome**?

- *A* What is the inheritance pattern for Aicardi syndrome?  
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- *I* What is the gender predilection?  
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There are case reports of Aicardi syndrome in phenotypical males with **47,XXY karyotype**

-- *R* Retardation

-- *D* Depigmentation  
**47,XXY males are syndromic. What is the eponymous name for this syndrome?**  
**Klinefelter's**

-- *I* Infantile seizures



'An x-linked recessive condition, lethal in genetically typical males such that all pts are female except for Klinefelter males.' There is only **one other condition** in the BCSC that fits this description—what is it?

what is the gender predilection?  
As it is lethal in males,  
--Coloboma of CNIII

the vast majority of pts are phenotypically female

'Vast majority'? If it's lethal in males, how do any survive to manifest the condition?  
There are case reports of Aicardi syndrome in phenotypical males with **47,XXY karyotype**

--Retardation

--Depig **47,XXY males are syndromic. What is the eponymous name for this syndrome?**  
**Klinefelter's**

--Infantile seizures

'An x-linked recessive condition, lethal in genetically typical males such that all pts are female except for Klinefelter males.' There is only **one other condition** in the BCSC that fits this description—what is it?  
Incontinentia pigmenti (IP)

What is the gender predilection?

As it is lethal in males, **the vast majority** of pts are phenotypically female

--Coloboma of CNII

'Vast majority'? If it's lethal in males, how do **any** survive to manifest the condition?

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*In one word, what sort of condition is IP?*

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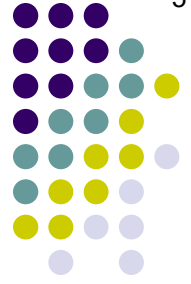
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## *Aicardi Syndrome*



Incontinentia pigmenti: Splashed-paint appearance

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**For more on IP (and the other phakomatoses), see slide-set P10**

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**And to be clear: For all this talk of Klinefelter's etc, if you are presented with an Aicardi syndrome pt on the OKAP or Boards, she will be female**

What is Aicardi syndrome?  
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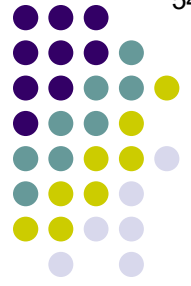
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## **Aicardi Syndrome**



*What findings comprise the Aicardi syndrome?*

*--Aicardi syndrome*

**Aicardi syndrome** is a rare disorder (<1:100,000 live births) that presents in females, usually during infancy. The first manifestation to be noted is often intractable seizures, usually infantile spasms. For such pts, DFE may be requested to assess for the presence of chorioretinal lacunae, which are considered pathognomonic. Colobomatous changes to the ONH are another common ocular manifestation (there are sporadic reports of myriad others). The classic finding on CNS imaging is absence or dysgenesis of the corpus callosum—but as with the eye, many other abnormalities have been reported. Facial dysmorphism can occur. The retardation is usually severe.

*--Depigmented chorioretinal lacunae*

*--Infantile seizures*

*Summary slide—no questions*