ANPH 252:

Skeletal Pathology CONGENITAL

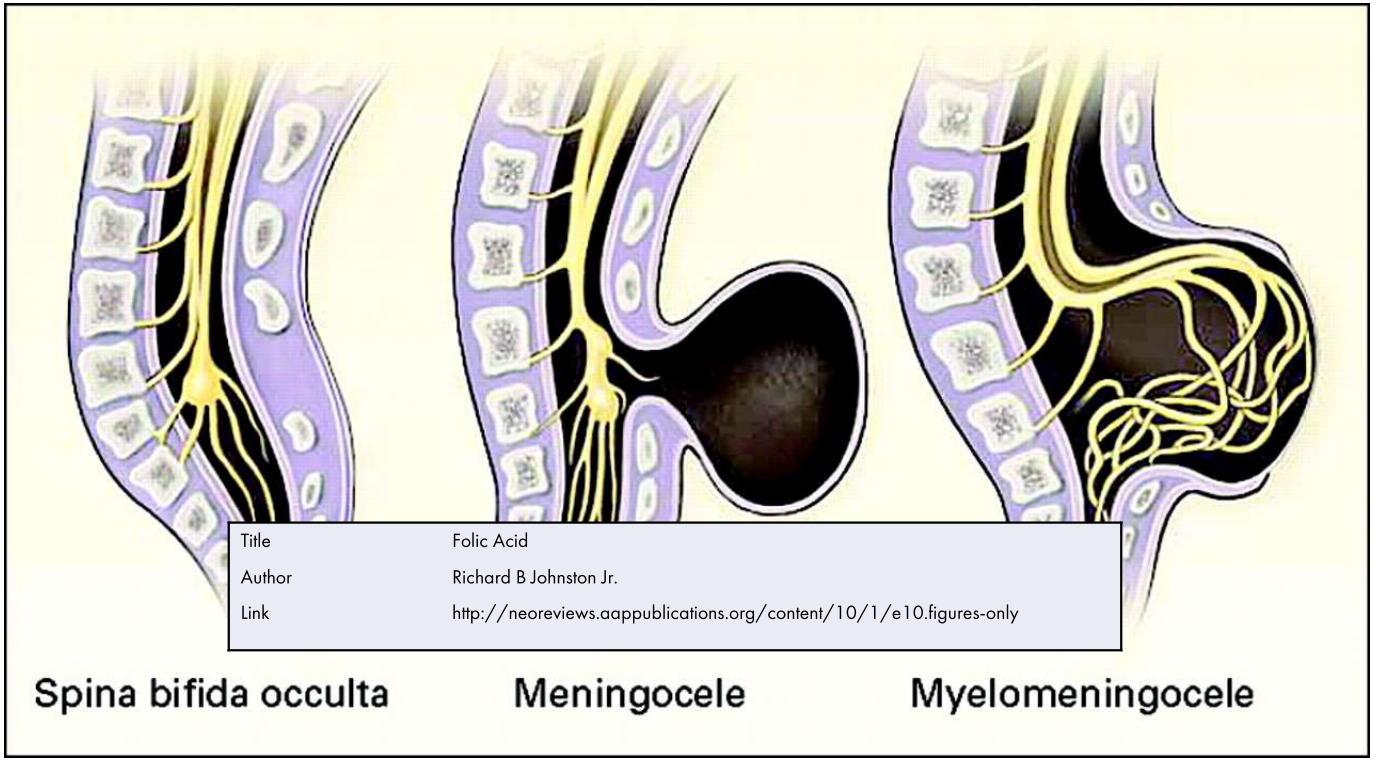
Congenital/Hereditary Diseases

- Spina Bifida
- Osteogenesis Imperfecta
- Osteopetrosis
- Congenital (developmental) Hip Dysplasia

Spina Bifida

- A posterior defect of the spinal canal
- Ranges in severity from having a bony split in the vertebral canal, to having a substantial absence of posterior bony structures, with herniation of the meninges and or spinal cord; spina bifida occulta, myelocele, meningocele, myelomeningocele (most common)
- Serious defects may cause spinal cord abnormalities inferior to the defect, and bladder and bowel incontinence
- Hydrocephalus is a common complication
- Surgical repair is required in severe cases
- Folic acid supplements taken during pregnancy will prevent this condition

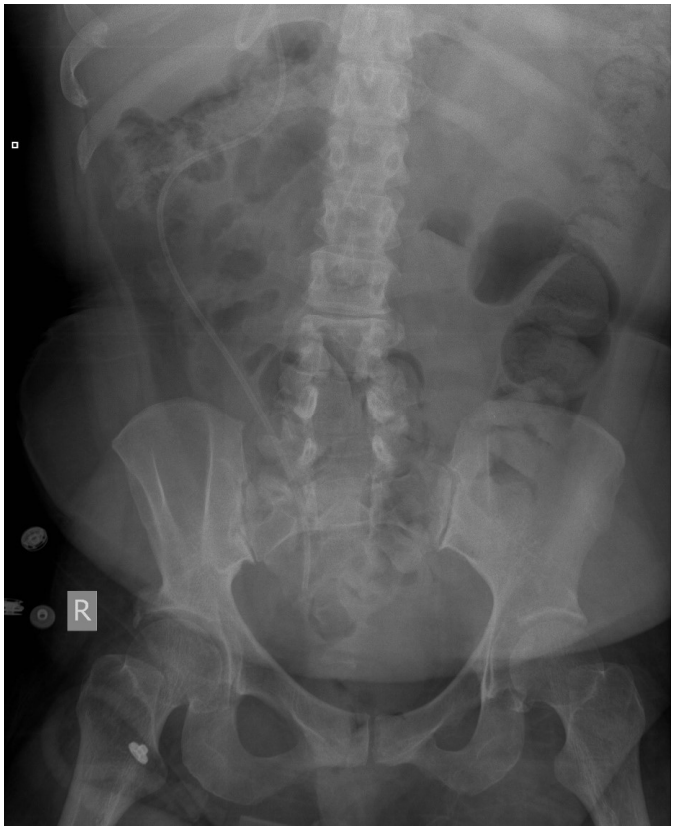
Spina Bifida



Title	Folic Acid
Author	Richard B Johnston Jr.
Link	http://neoreviews.aappublications.org/content/10/1/e10.figures-only


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Eisenberg (6th ed.) fig. 4-4
Spina bifida occulta: insignificant split in the bony canal



Case courtesy of Dr Ian Bickle, Radiopaedia.org, rID: 45935

Osteogenesis Imperfecta "brittle bone disease"

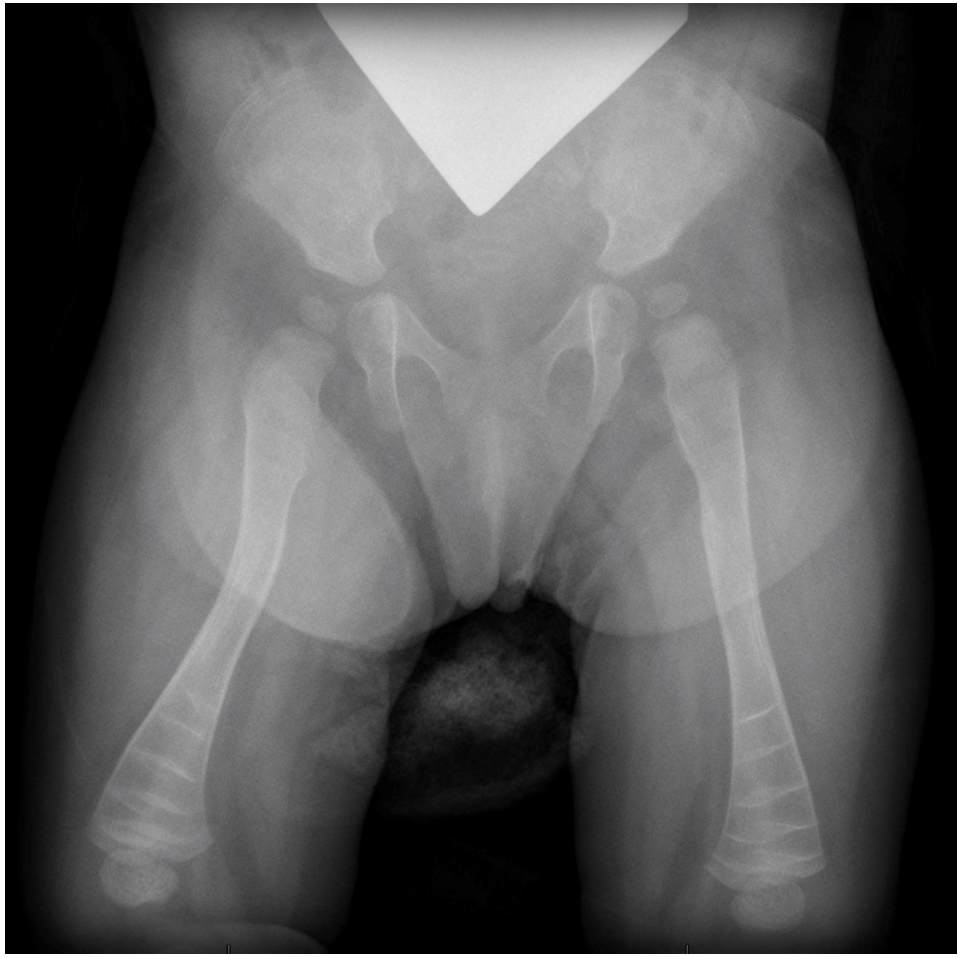
- Connective tissue disorder characterized by osteoporosis with abnormal bone fragility, hearing impairment, dentinogenesis imperfecta and blue sclera

- Tend to be wheelchair bound as the bones cannot support body weight

- Use caution when moving and positioning these patients, and consider decreasing kV


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Eisenberg Fig 4-7



Case courtesy of Dr Michael Sargent, Radiopaedia.org, rID: 6037

"Zebra stripe sign is seen in children with OI who have been treated with cyclical bisphosphonate therapy. When the drug is delivered in cycles, dense bone is formed while treatment is being given. This results in dense stripes across the metaphyses of bones which can be visualized radiographically."

Osteogenesis imperfecta



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DescriptionCharacteristically blue sclerae of patient with osteogenesis imperfecta.jpg	**English:** The classic blue sclera of a person with osteogenesis imperfecta
Date	
Source	http://cnx.org/contents/_on798ZB@3/Images-of-Memorable-Cases-Case
Author	Herbert L. Fred, MD and Hendrik A. van Dijk

Osteogenesis imperfecta





Eisenberg (6th ed.) Fig 4-8 A: multiple fractures of ribs and long bones. Overly exuberant callus formation can mimic tumours, and cause bizzare deformities. May be confused with child abuse
B: Skull bones fuse slowly and wormian bones develop to close the gaps

Osteopetrosis

- "Marble bone disease"
- Rare hereditary disorder
- Caused by defective osteoclasts
- Prevents bone marrow from forming
- Bones are sclerotic and thick, but are very weak

Osteopetrosis



Case cou Maulik S Patel, Radiopaedia.org, rID: 9142

Developmental Dysplasia of the Hip

- Incomplete formation of the acetabulum
- During pediatric assessment the hip might "pop" out when abducting or flexing the leg
- AP and bilateral frog leg (modified Cleaves) will help diagnose; femur will be displaced superiorly and posteriorly.
- Casting or splinting will allow the acetabulum to grow

Developmental Dysplasia of the Hip

- If the child is already walking, they may waddle like a duck



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Fig 4-12 – Eisenberg (6th ed.)

DDH



Title	Quick DDH Update
Author	Flunderbunk in DDH
Link	https://flunderbunk.files.wordpress.com/2013/05/2012-11-16-12-40-43.jpg

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