



The Great Mimicker: A Misdiagnosis of Child Abuse

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No financial disclosures

Outline:

- Case presentation
- Discussion about child physical abuse
- Discussion about the confirmed diagnosis

Case Presentation:

I was consulted on 9 mo twins (boy and girl) due to abnormal blood test

But I must start from the very beginning

Birth and prenatal histories:

- Infants born at 37 wks of gestation by C-section due to malpresentation
- No major perinatal complications
- Mom was 24 yo G2P1A1L0→2
- Negative for GBS, gonorrhea, Chlamydia, Trichomonas, HIV, RPR and hepatitis B
- Rubella immune, parvovirus B19 unknown
- Urine drug screen x2 negative
- Apgar scores 8 and 9 at 1 min and 5 min, respectively
- Passed hearing, newborn and critical congenital heart disease screens
- Discharged home at day 3 of life

Clinical Course:

-At 3 wks of age, boy started to have fussiness/crying, decreased finger grip and decreased wrist motion b/l

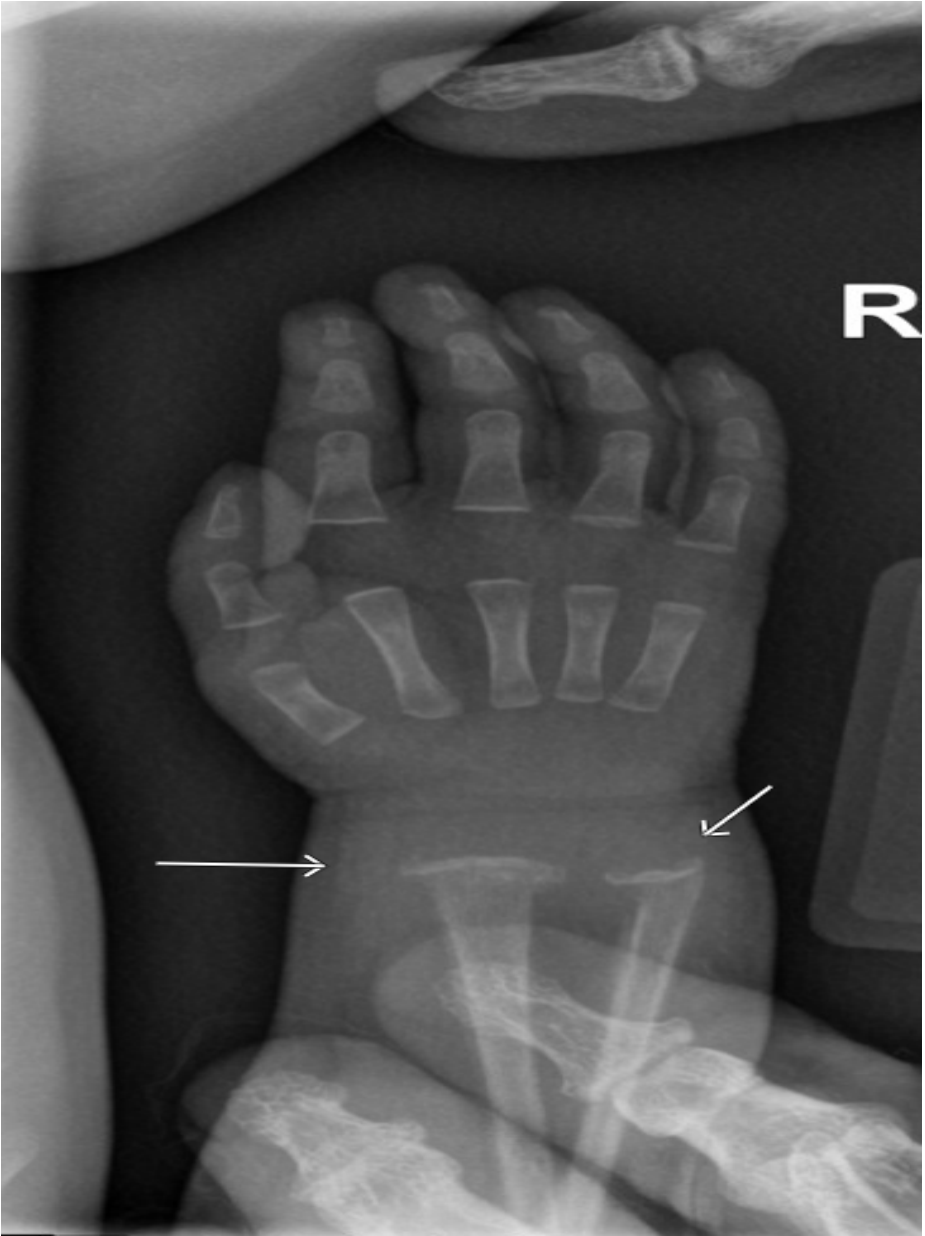
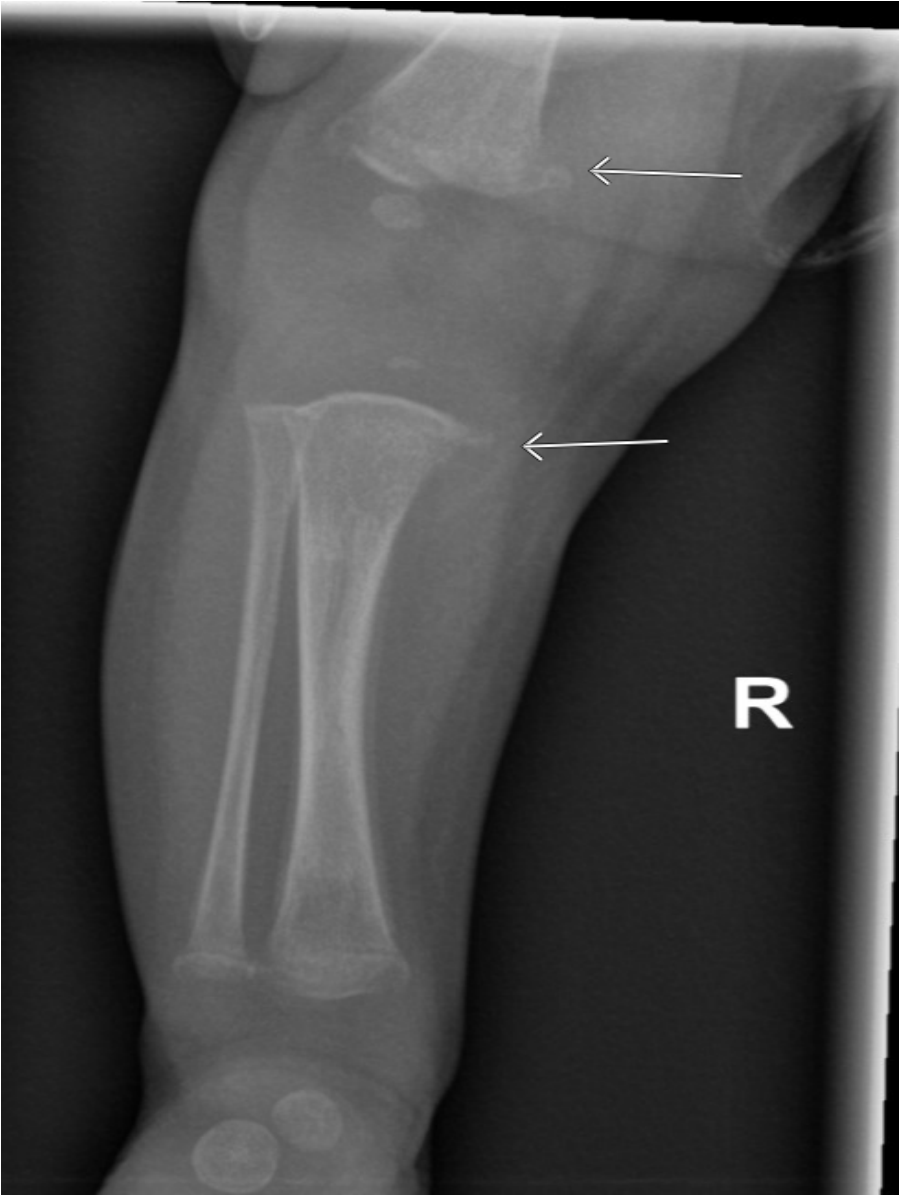
-Physical exam at 5 wks of age: decreased finger grip but moving all extremities

-Skeletal series showed:

Fractures in bilateral distal femoral, proximal tibial, distal radius and ulnar metaphyses

Some may be healing and are associated with periosteal reaction

Boy

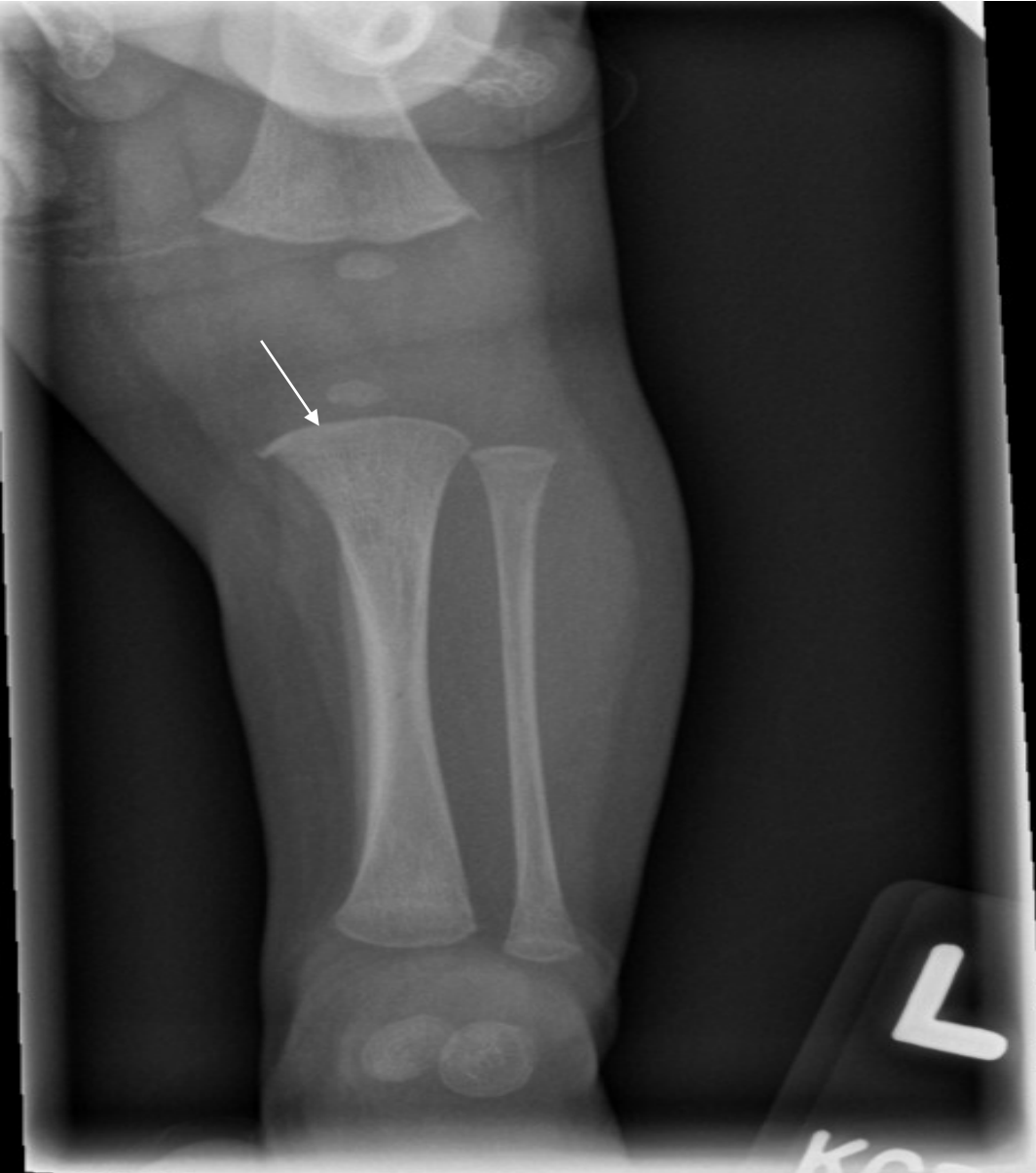


Clinical Course:

-Sister was asymptomatic but had skeletal series due to the findings seen in her brother

-X-rays suspicious for periosteal new bone in tibial metaphysis b/l and osteophytes in distal femur b/l

Girl



Clinical Course:

- Both infants then admitted due to concerns of NAT
- No evidence of head or neck trauma on further imaging
- DHHS involved
- Discharged with foster family due to concerns for child physical abuse

Child Physical Abuse:

-Fractures are the most common injury caused by child physical abuse after bruises

-1/4 of fractures in children <1 year are caused by child abuse

-However, incorrectly diagnosing physical abuse in child with noninflicted fractures has serious consequences for child and family

Differential Diagnosis of Fractures in Infants:

- Trauma: accidental or abusive
- Birth injury
- Osteogenesis imperfecta
- Vitamin D deficiency rickets
- Scurvy
- Osteopenia of prematurity
- Demineralization from disuse
- Osteomyelitis
- Copper deficiency or Menkes disease
- Systemic disease

chronic renal/liver disease, Fanconi syndrome, hypophosphatasia, hyperparathyroidism, RTA

Fractures Concerning for Child Abuse:

- No history of injury
- Fractures not consistent with history/mechanism
- Inconsistent histories
- Fracture mechanism not consistent with child's age and development
- Delay in obtaining medical treatment
- Fractures with high specificity for abuse
- Multiple fractures
- Fractures of different ages or stages of healing
- Presence of other injuries suspicious for abuse (e.g., injuries to skin, internal organs or CNS)

TABLE 1 Specificity of radiologic findings in infants and toddlers¹⁹

High specificity^a

CMLs

Rib fractures, especially posteromedial

Scapular fractures

Spinous process fractures

Sternal fractures

Moderate specificity

Multiple fractures, especially bilateral

Fractures of different ages

Epiphyseal separations

Vertebral body fractures and subluxations

Digital fractures

Complex skull fractures

Common, but low specificity

Subperiosteal new bone formation

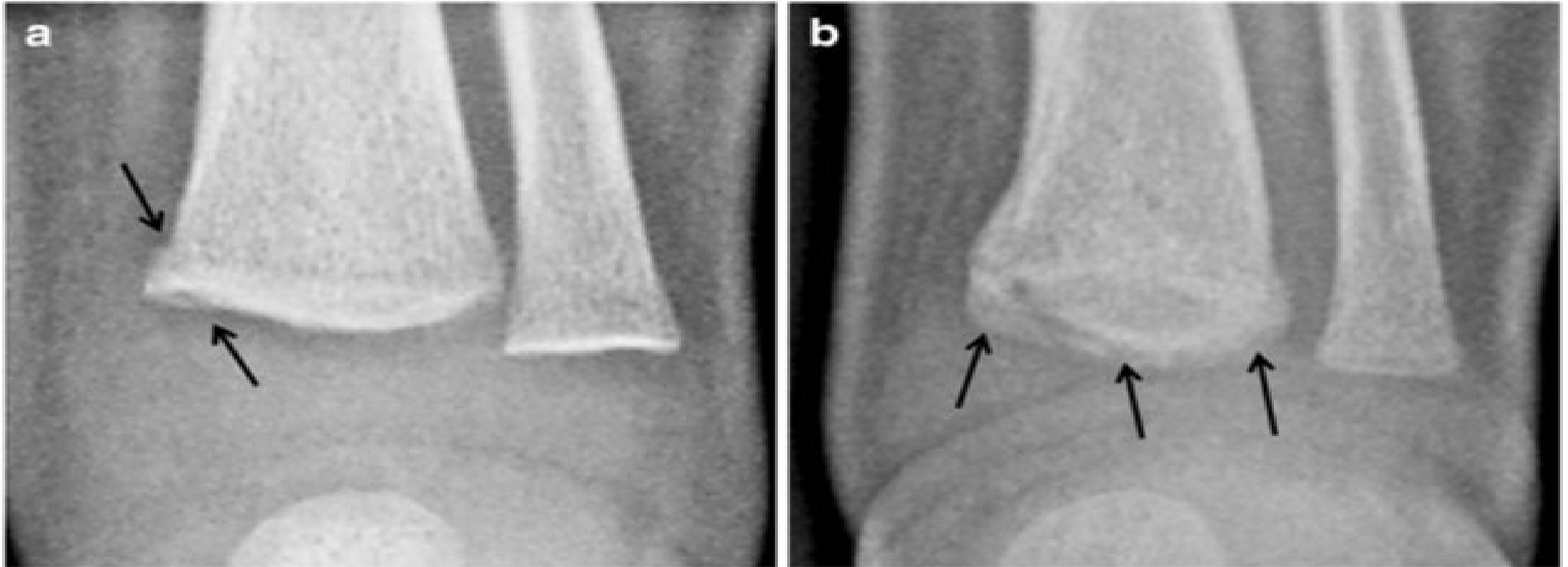
Clavicular fractures

Long-bone shaft fractures

Linear skull fractures

^a Highest specificity applies in infants.

Classic metaphyseal lesions (CMLs) are the most common long bone fracture found in infants who die with evidence of inflicted injury



Back to Clinical Course:

- Evaluated by Genetics: negative osteogenesis imperfecta and bone fragility panel
- Evaluated by Pediatric Endocrinology: negative testing for rickets, hypophosphatasia or hypoparathyroidism
- Child abuse diagnosis became more obvious (multiple long-bone fractures with metaphyseal involvement)

Clinical Course:

- Boy noted to have nasal congestion and noisy breathing since arrival to foster family
- Evaluated by ENT: no evidence of choanal atresia
- Had b/l nasal stents placed at 5 mo of age with significant improvement
- Had failure to thrive, needing temporary NG-tube feeding
- Had gross motor developmental delays, needing PT

Clinical Course:

- At 1 wk of age, girl started having nasal congestion/obstruction leading to noisy breathing and poor feeds
- Evaluated by ENT: no evidence of choanal atresia
- Had b/l nasal stents placed at 2.5 mo of age with no significant improvement
- Had failure to thrive, needing temporary NG-tube feeding
- Had perianal ulcers at 8.5 mo of age

Findings' Summary:

- Multiple symmetrical fractures
- Chronic nasal congestion/obstruction
- Failure to thrive
- Gross motor delays (boy)
- Perianal ulcers (girl)

More About Clinical Course:

- When infants were 8.5 mo, biological mom found to have reactive syphilis test upon plasma donation
- This was confirmed at PCP's office → RPR 1:8 with reactive FTA IgG
- Negative for HIV, gonorrhea, Chlamydia and Trichomonas

Clinical Course:

- Maine CDC notified about mom's postnatal syphilis and found out that she did not have prenatal syphilis test
- An old RRP result was scanned with a wrong date and looked like it was done during first trimester
- PCP asked to check infants' RPR

Clinical Course:

At 9 mo of age:

-RPR → boy: 1:32

→ girl: 1:512

-Both infants admitted

-Physical exam → boy: central hypotonia

→ girl: noisy breathing and small perianal ulcers

-Labs → boy: normal WBC, reactive lymph=10%, Hb=9.9, MCV=69.7, normal platelets, normal LFTs/bilirubin

→ girl: normal WBC, reactive lymph=2%, Hb=9.1, MCV=69.6, normal platelets, normal LFTs/bilirubin

Clinical Course:

-CSF

→ boy: 104 WBCs, 34 RBCs, 87% lymph, glucose 44, ptn 48, VDRL 1:16

→ girl: 3 WBCs, 13 RBCs, neut 23%, lymph 39%, mono 38%, glucose 56, ptn 25, VDRL 1:2

-Normal MRI brain and eye/hearing exams

-Repeat skeletal series: healing fractures, stable/residual periosteal reaction, no new fractures

-Negative for gonorrhea, Chlamydia, Trichomonas, Hep B and Hep C

-Treated with IV penicillin G 50,000 units/kg/dose q6 hours for 14 days

Clinical Course:

- Biological mom admitted later that she was a sex worker in NY for couple years
- This ended in Oct 2021
- Not shared with Ob
- Supposedly got pregnant in Dec 2021
- HealthInfoNet showed that RPR was collected in outside lab on 1/27/21 but was added to mom's chart with the date of 1/27/22

Clinical Course:

-Infants back to their biological mother since NAT was thought to be less likely

-RPR: admission → 2.5 mo → 6 mo

Boy: 1:32 → 1:16 → 1:8

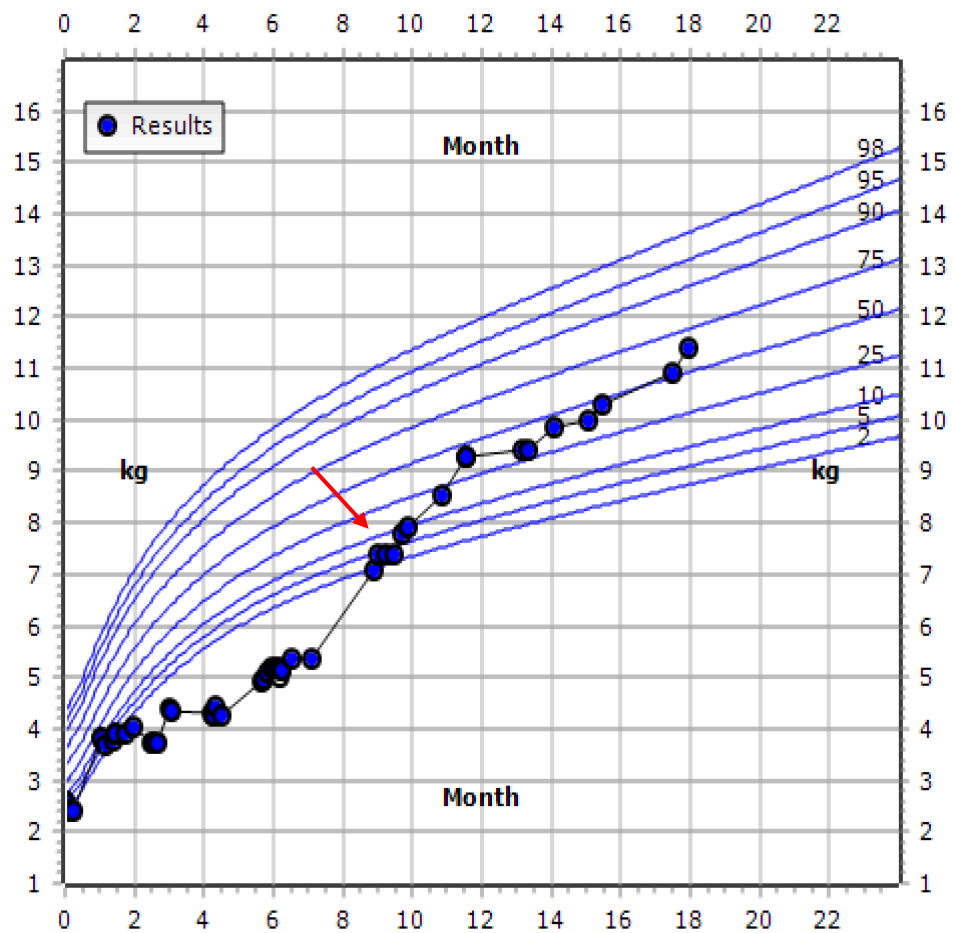
Girl: 1:512 → 1:256 → 1:32

-Repeat CSF 6 mo later showed resolution of neurosyphilis

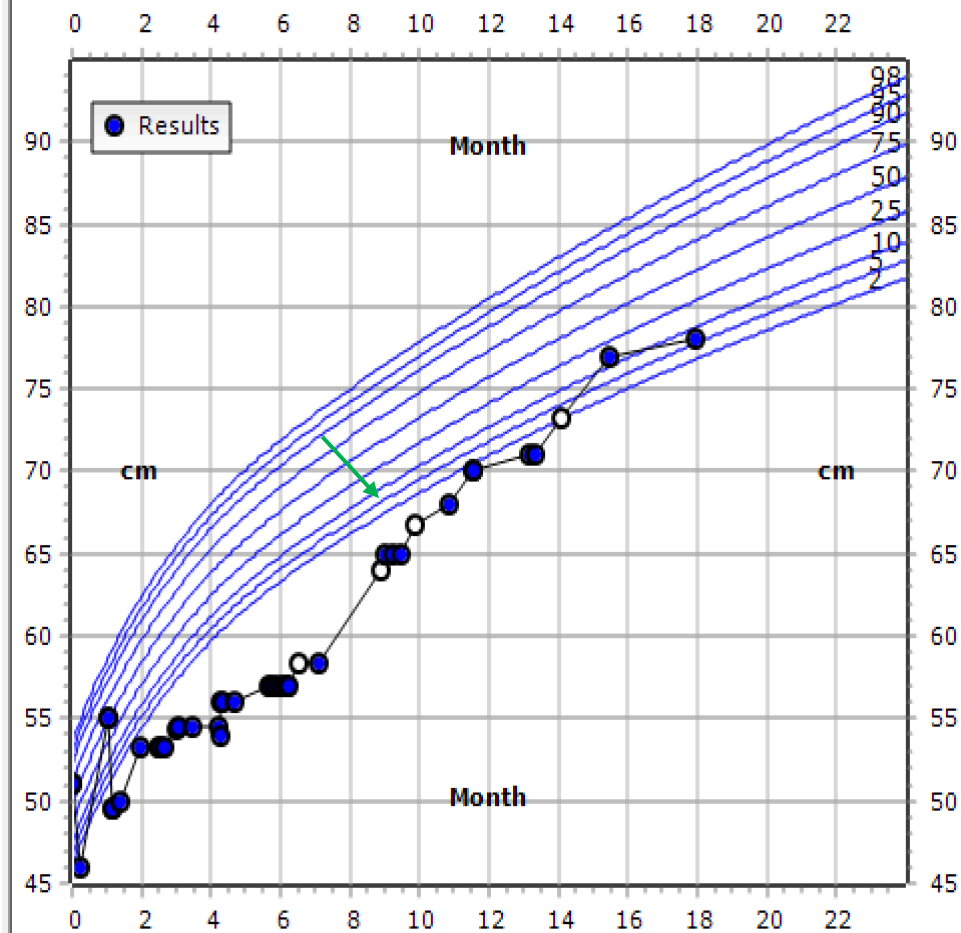
-Growing well

-Mild developmental and speech delays

CDC's WHO Weight-for-age, 0-2 years, Boys



CDC's WHO Length-for-age, 0-2 years, Boys



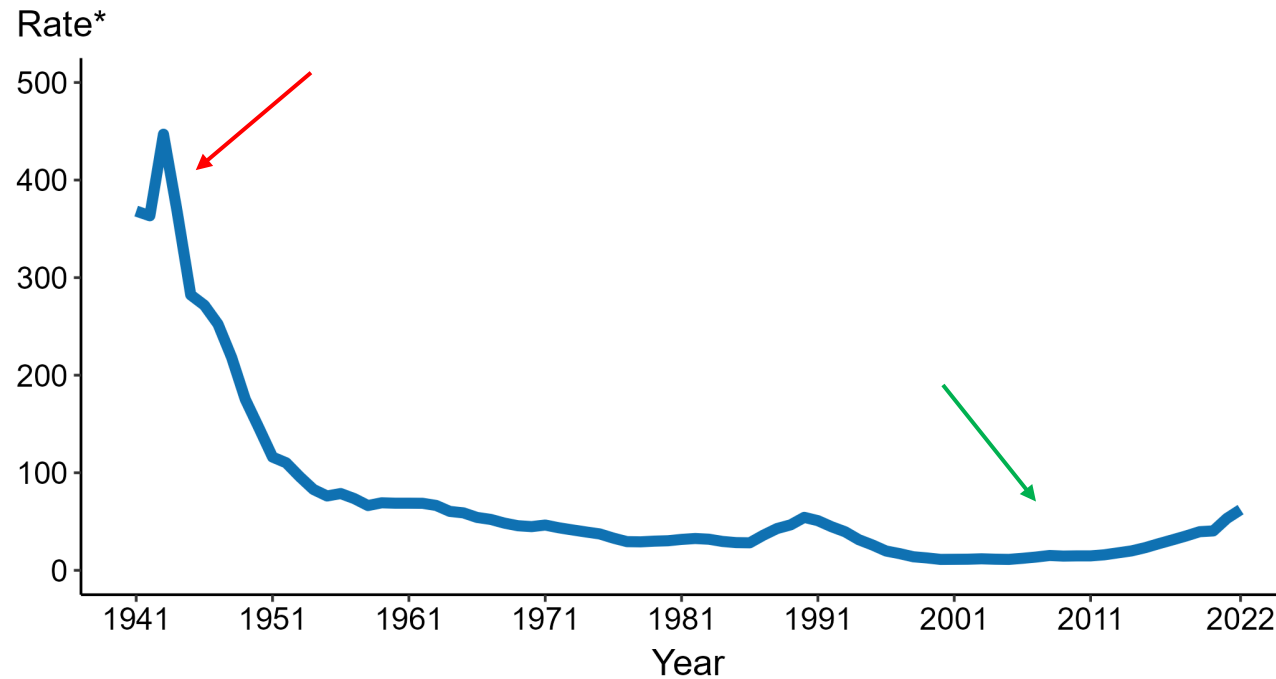
Syphilis:

Caused by the spirochete *Treponema pallidum*





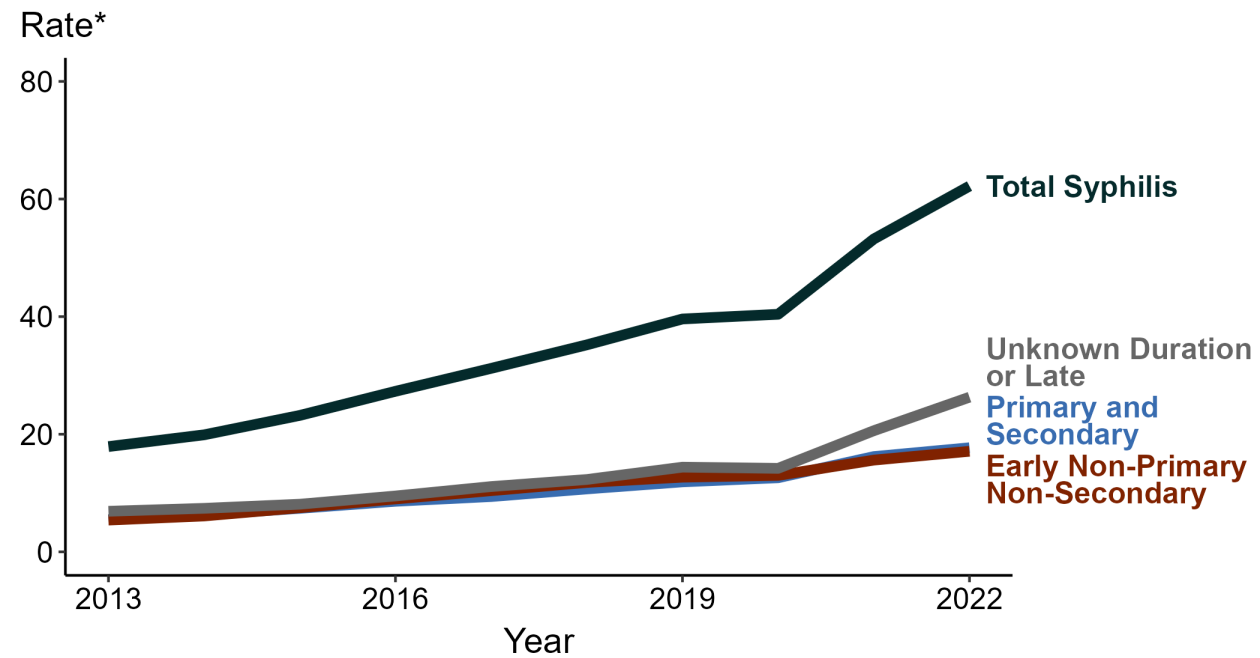
Syphilis — Rates of Reported Cases (All Stages) by Year, United States, 1941–2022



* Per 100,000



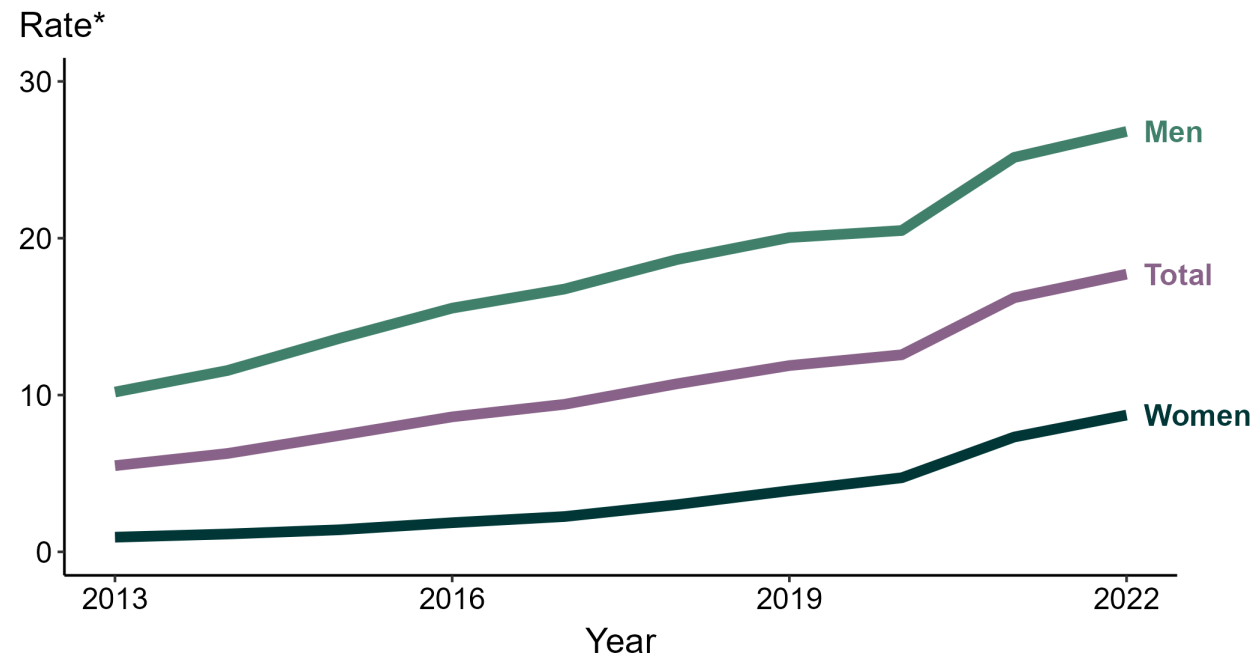
Syphilis — Rates of Reported Cases by Stage of Infection, United States, 2013–2022



* Per 100,000



Primary and Secondary Syphilis — Rates of Reported Cases by Sex, United States, 2013–2022



* Per 100,000

Statistics in Maine:

-From 2012-2021, syphilis cases increased from 20 to 101 (405%) per year



Case Information:

101

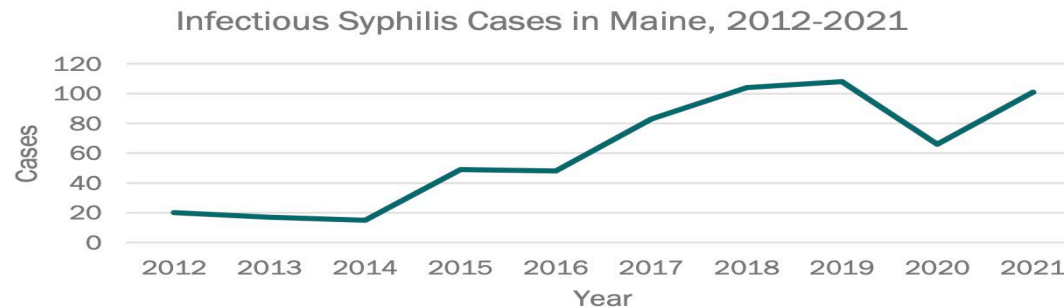
Cases of infectious syphilis

7.4

Cases per 100,000 people

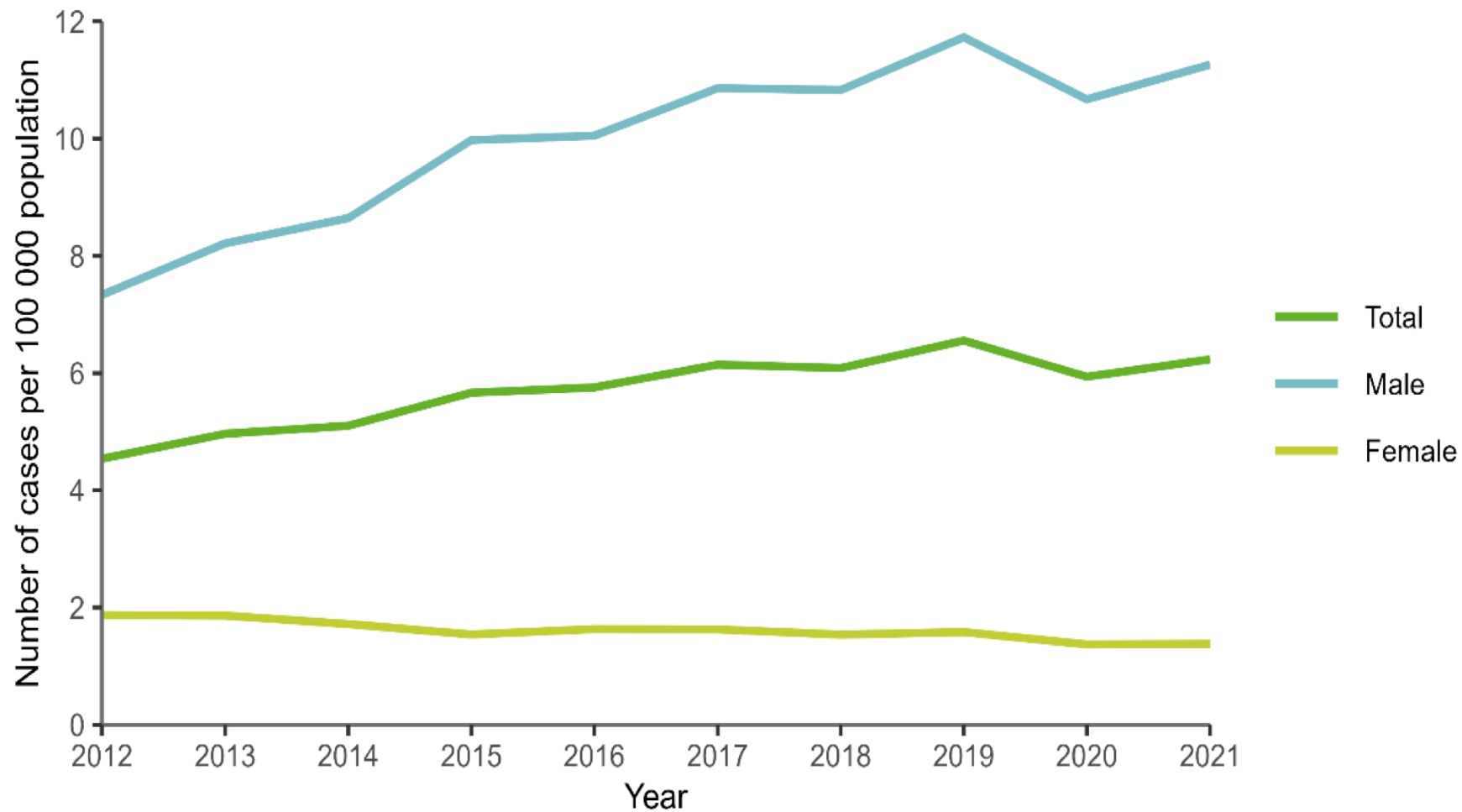
405%

Increase since 2012



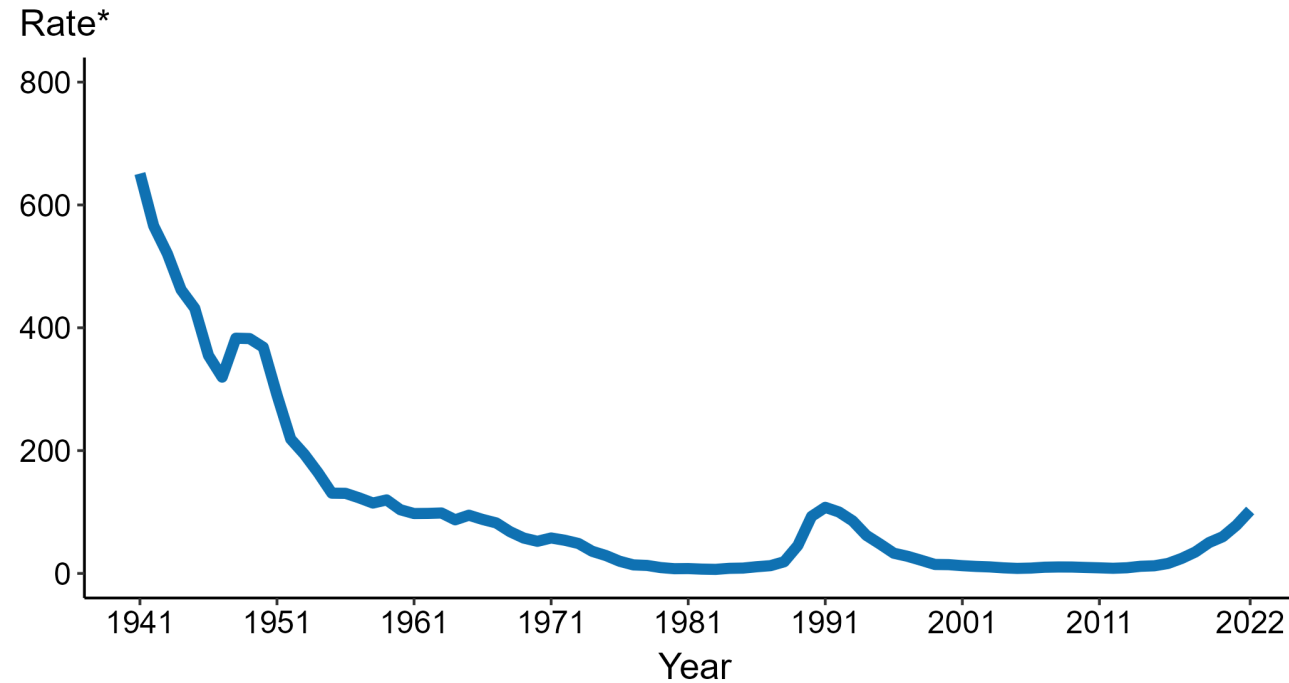
-In 2022, prelim data show that, of the 112 reported cases of syphilis in Maine, 20.5% were among women 15-44 years

Figure 4. Rate of confirmed syphilis cases per 100 000 population, total and by gender for cases with available data, EU/EEA countries reporting consistently, 2012–2021





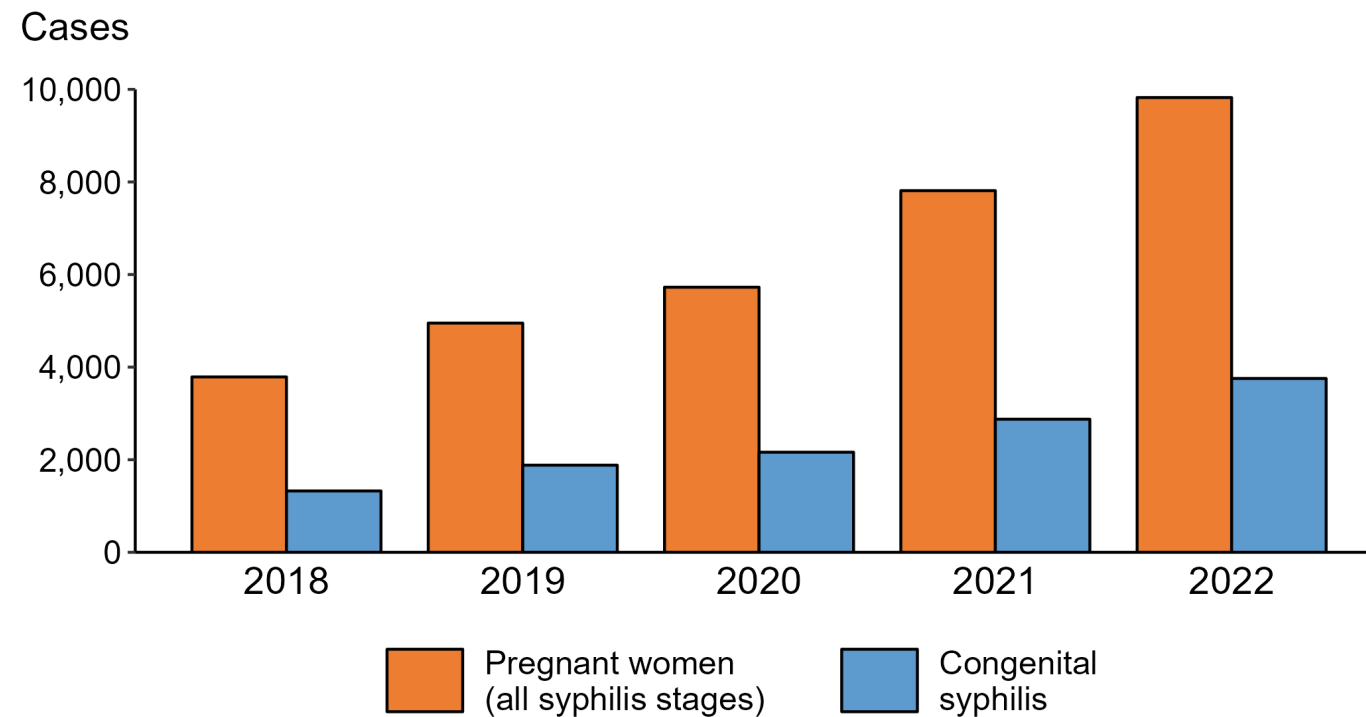
Congenital Syphilis — Rates of Reported Cases by Year of Birth, United States, 1941–2022



* Per 100,000 live births

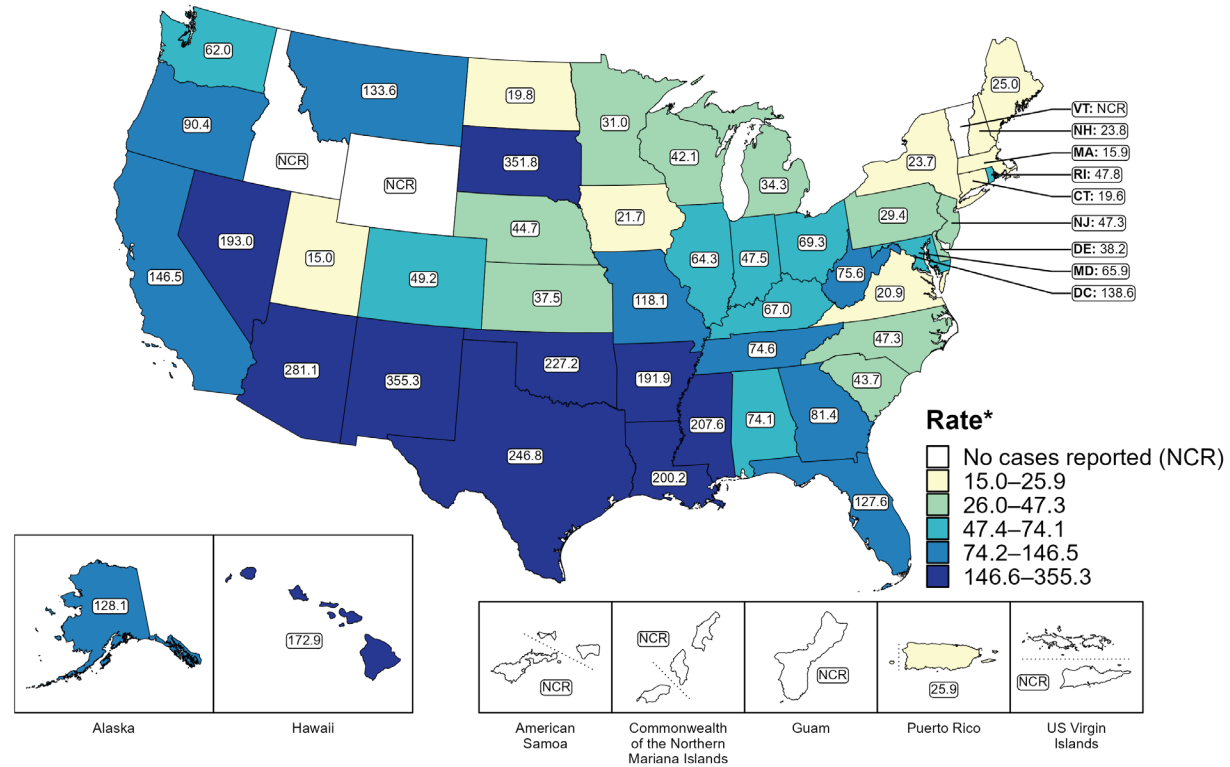


Syphilis— Reported Cases of Syphilis (All Stages) among Pregnant Women and Reported Cases of Congenital Syphilis by Year of Birth, United States, 2018–2022





Congenital Syphilis — Rates of Reported Cases by Jurisdiction, United States and Territories, 2022



* Per 100,000 live births



Congenital Syphilis — Reported Cases by Year of Birth and Jurisdiction, United States and Territories, 2013 and 2022

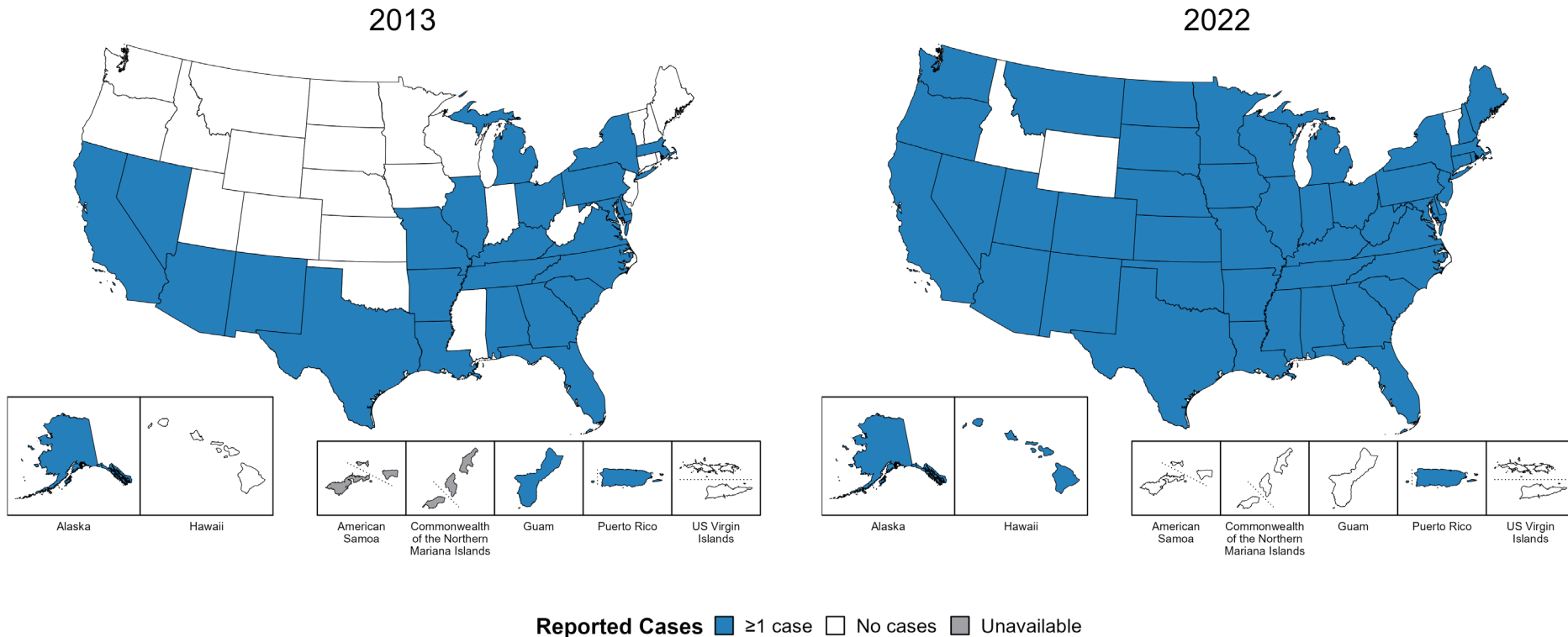




Table 31. Congenital Syphilis — Reported Cases and Rates of Report...
Cases by State, Ranked by Rates, United States, 2022

Rank*	State†	Cases	Rate per 100,000 Live Births
1	New Mexico	76	355.3
2	South Dakota	40	351.8
3	Arizona	219	281.1
4	Texas	922	246.8
5	Oklahoma	110	227.2
6	Mississippi	73	207.6
7	Louisiana	115	200.2
8	Nevada	65	193.0
9	Arkansas	69	191.9
10	Hawaii	27	172.9
11	California	616	146.5
12	Montana	15	133.6
13	Alaska	12	128.1



Table 31. Congenital Syphilis — Reported Cases and Rates of Reported Cases by State, Ranked by Rates, United States, 2022

37	Minnesota	20	31.0
38	Pennsylvania	39	29.4
39	Maine	3	25.0
40	New Hampshire	3	23.8
41	New York	50	23.7
42	Iowa	8	21.7
43	Virginia	20	20.9
44	North Dakota	2	19.8
45	Connecticut	7	19.6
46	Massachusetts	11	15.9
47	Utah	7	15.0
	Idaho	0	0.0
	Vermont	0	0.0
	Wyoming	0	0.0

Prenatal Syphilis Testing:

-Rapid plasma reagin (RPR) should be obtained **early in pregnancy**

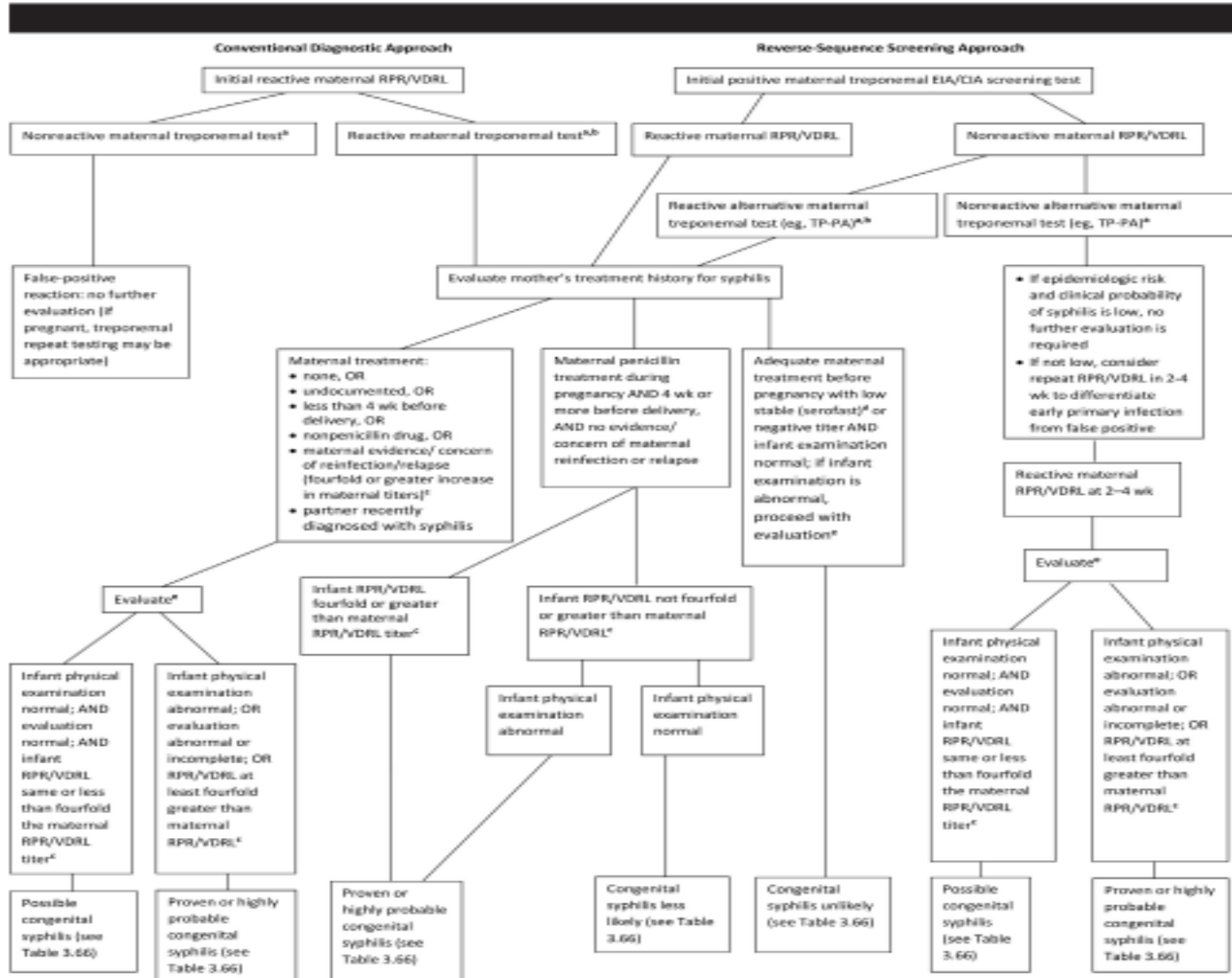


-Pregnant women should be retested at **28 weeks'** gestation and **at delivery** if the mother lives in a community with high syphilis rates or is at risk for syphilis acquisition during pregnancy (e.g., misuses drugs or has an STI during pregnancy, having multiple sex partners, having a new sex partner, or having a sex partner with an STI)

-Neonates should not be discharged from the hospital unless the syphilis status of the mother has been determined at least once during pregnancy

-Any woman who delivers a stillborn infant should be tested for syphilis

FIG 3.15. ALGORITHM FOR DIAGNOSTIC APPROACH OF INFANTS BORN TO MOTHERS WITH REACTIVE SEROLOGIC TESTS FOR SYPHILIS.



Congenital syphilis:

-Ancient disease

-May be contracted at any stage of maternal infection via transplacental transmission at any time during pregnancy or via contact with maternal lesions at time of delivery¹

-Adequate maternal treatment >4 weeks prior to delivery prevents **up to 98%** of congenital syphilis cases²

1. Red Book: 2021–2024 Report of the Committee on Infectious Diseases. By: Committee on Infectious Diseases, American Academy of Pediatrics, David W. Kimberlin, MD, FAAP, Elizabeth D. Barnett, MD, FAAP, Ruth Lynfield, MD, FAAP, Mark H. Sawyer, MD, FAAP

2. Jacobs K, Vu DM, Mony V, Sofos E, Buzi N. Congenital Syphilis Misdiagnosed as Suspected Nonaccidental Trauma. Pediatrics. 2019 Oct;144(4):e20191564. doi: 10.1542/peds.2019-1564. Epub 2019 Sep 19. PMID: 31537633.

Congenital Syphilis:

Presentation:

- Stillbirth
- Hydrops fetalis
- Preterm birth
- HSM
- Snuffles
- Lymphadenopathy
- Mucocutaneous lesions
- Maculopapular rash mostly on hands and feet
- Pneumonia
- Osteochondritis
- Periostitis
- Pseudoparalysis
- Anemia
- Thrombocytopenia
- Jaundice
- Asymptomatic



Congenital Syphilis Misdiagnosed as Suspected Nonaccidental Trauma

Kimberley Jacobs, MD,^a David M. Vu, MD,^{a,b,c} Vidya Mony, MD,^b Elvera Sofos, MD,^b Nadav Buzi, MD^b


PEDIATRICS Volume 144, number 4, October 2019:20191564

Pediatric Imaging Case Report

Congenital syphilis, the great mimicker, confused with non-accidental trauma

Preet Kiran Sandhu¹

Congenital syphilis as the cause of multiple bone fractures in a young infant case report

Maria Koliou^{1,2*} , Elpida Chatzicharalampous², Myria Charalambous² and Kyriakos Aristeidou³

Koliou *et al. BMC Pediatrics* (2022) 22:728

Congenital Syphilis:

- Skeletal manifestations in 60-80% of infants born to mothers with untreated congenital syphilis
- Rare cause of long-bone fractures
- Long-bone osteoperiostitis → pseudoparalysis and fractures
- Prompts workup for NAT and delays evaluation for congenital syphilis

Congenital Syphilis:

-Skeletal involvement is usually bilateral, symmetrical and diffuse

-The most common skeletal lesions are diaphysitis and metaphysis

Table III. Radiological lesions

	n=197 Number of cases
Diaphysitis	126
<i>Periosteal reaction</i>	106
Mild	72
Severe (lamellar)	34
<i>Osteitis</i>	20
Localised defect	5
Diffuse	15
Metaphysitis	71
Dense bands	10
Lucent bands	15
Alternating bands	12
Metaphyseal serrations	4
Exuberant callus formation	3
Wimberger's sign	21
Focal changes	6
Combined lesions	61
Asymmetrical lesions	15
Dactylitis	4
Pathological fractures	12
Joint involvement	8

Metaphyseal lesions due to congenital syphilis in a 4 mo girl



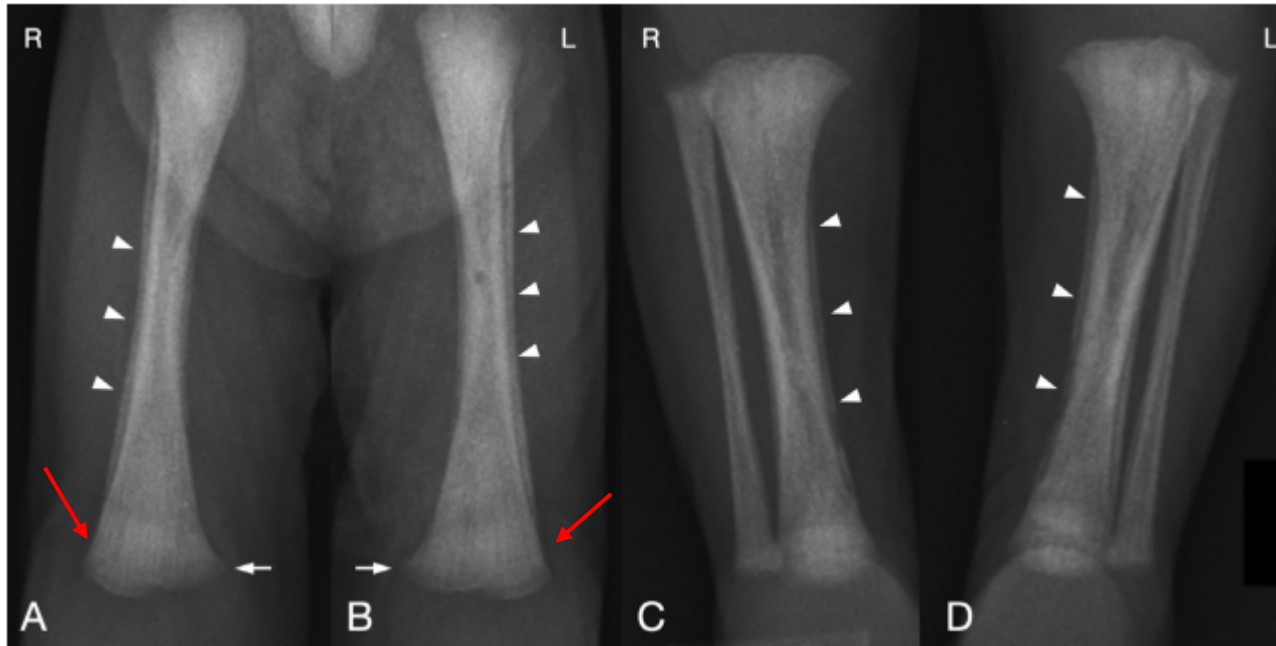
Metaphyseal lesion due to congenital syphilis in the same 4 mo girl



Wimberger sign:

Metaphyseal destruction of the proximal medial tibia

Symmetrical bony lesions in a preterm neonate



Arrowheads: periosteal reactions
White arrows: subtle lucencies
Red arrows: "celery stalk" appearance

Congenital Syphilis:

If untreated, may lead to late manifestations usually >2 years of age:

- Interstitial keratitis
- CN8 palsy
- Deafness
- Hutchinson teeth
- Anterior bowing of shins
- Frontal bossing
- Saddle nose
- Rhagades



Painting of 16 yo girl with late congenital syphilis (1875-1882)



Congenital Syphilis:

Diagnosis:

In addition to mom's prenatal syphilis testing, treatment and time of treatment, we rely on infant's:

- Physical exam
- Nontreponemal test (e.g., RPR or VDRL)
- CSF cell count, protein and VDRL
- CBC/diff
- LFTs, long-bone X-rays, chest X-ray, brain imaging, eye exam, and ABR

Congenital Syphilis:

Diagnosis:

-Not recommended to check treponemal tests due to passive transmaternal transfer until 15 mo of age

-Category: **Proven or Highly probable** vs. **Possible** vs. **Less likely** vs. **Unlikely**

Congenital Syphilis:

Treatment:

-Penicillin G

-Duration depends on likelihood of of congenital syphilis



Congenital Syphilis:

Monitoring:

- Nontreponemal tests should be performed every 2-3 mo until nonreactive
- Typically decrease by 3 mo and become nonreactive by 6 mo
- May have slower response in infants treated after the neonatal period
- No need to repeat CSF after 6 mo in neonates who have neurosyphilis if titers are trending down

Congenital Syphilis:

Late sequelae:

-Limited reports in the literature

-In a 5-year follow-up study, 16/120 (13%) infants treated for congenital syphilis had sequelae

-14/16 children had developmental delays

-14/16 children had other sequelae: hydrocephalus, microcephaly, cortical atrophy, GH deficiency, severe anemia, etc

Conclusion:

- We should be aware of the rising rates of syphilis and congenital syphilis
- All pregnant women should be screened for syphilis early in pregnancy then retested in third trimester if high risk
- Long-bone fractures in infants should always prompt consideration of child abuse
- However, other causes of fractures should be entertained
- Consider congenital syphilis in the differential diagnosis of infants with symmetrical long-bone fractures, especially in case of other signs/symptoms
- Proper documentation of outside results

