



Women's
& Children's
Hospital
ADELAIDE



MP Metz



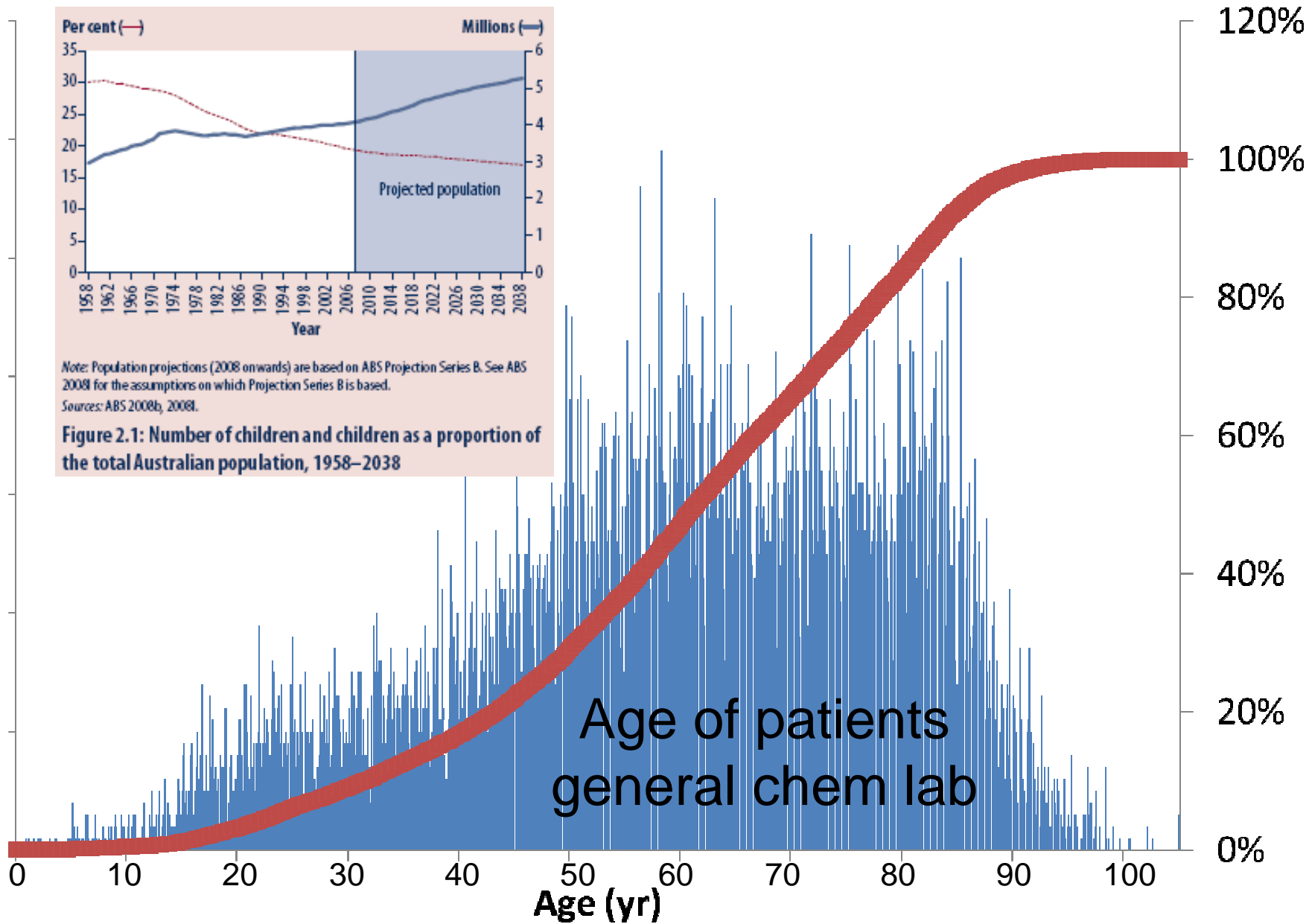
Cases in Kids' Chemistry



National Scientific Meeting
Medical Science - **Today and Tomorrow**

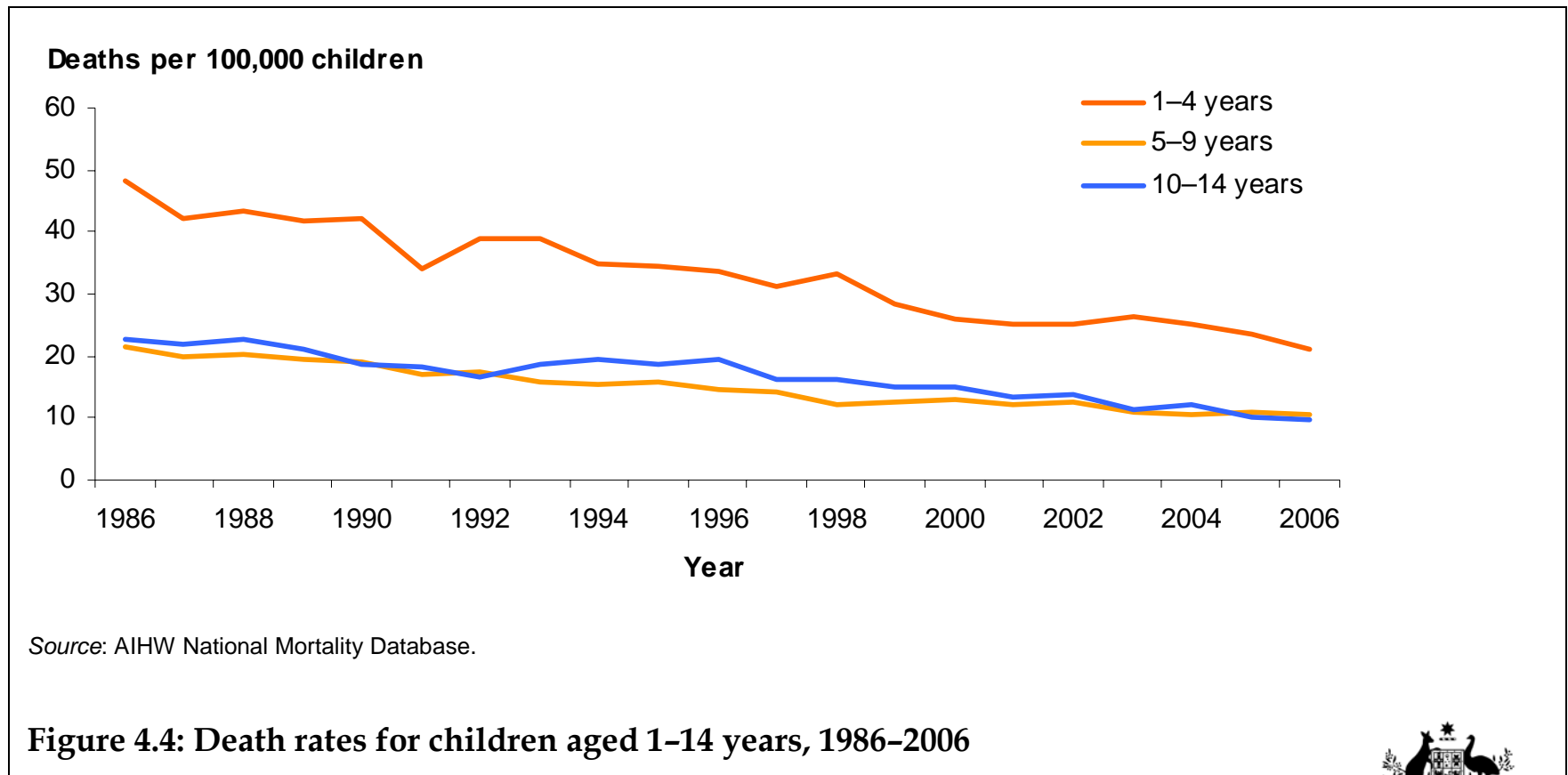


Adelaide 12-16 October 2009 | Adelaide Convention Centre, South Australia



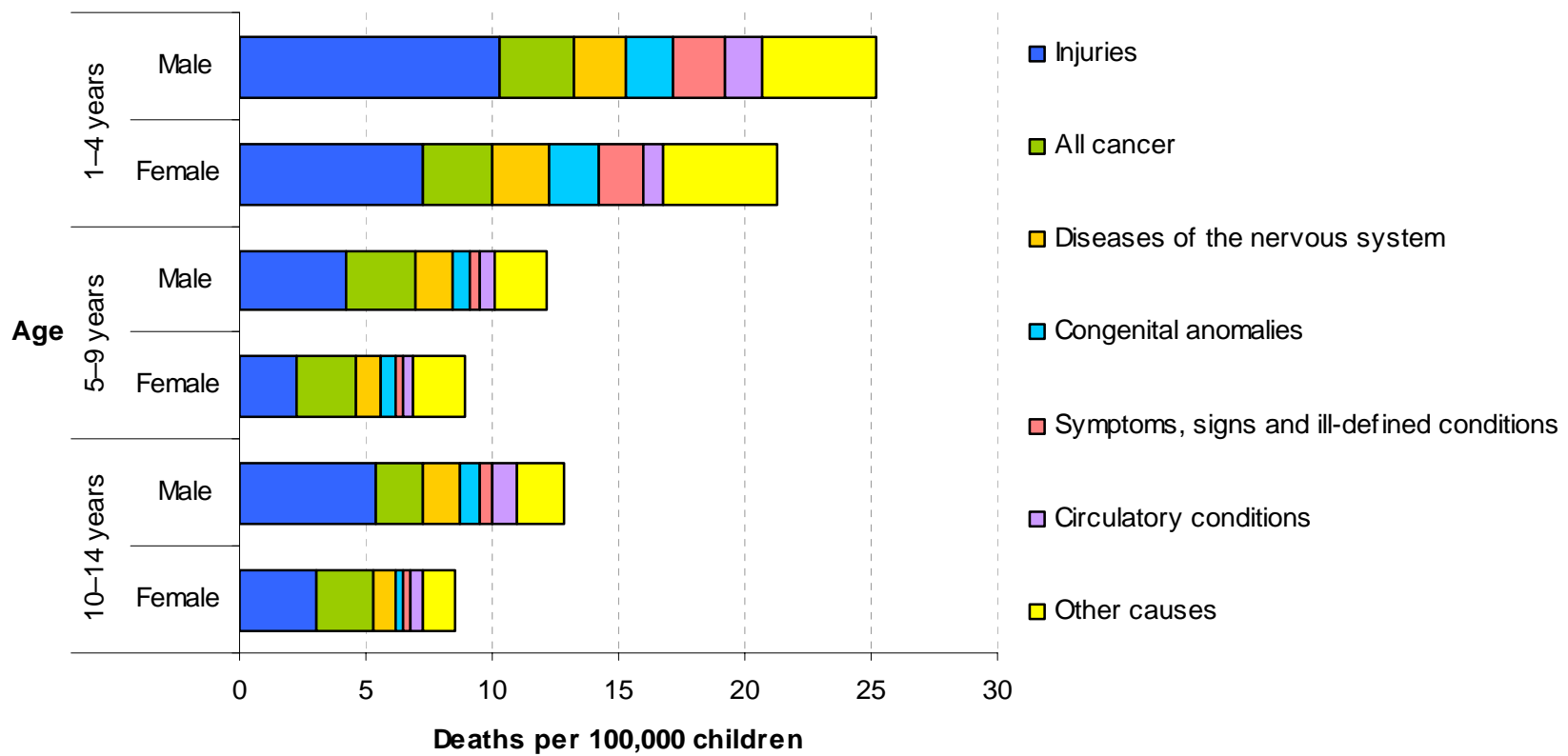
Child aged 5-9 = 1/2000

Adult aged 50-54 = 1/71



Australian Government

Australian Institute of
Health and Welfare



Notes

1. Refer to Table A1.2 for ICD-10 codes.
2. Other causes accounted for 17% of child deaths among 1-14 year olds in 2004-2006.

Source: AIHW National Mortality Database.

Figure 4.5: Leading causes of death among children aged 1-14 years, 2004-2006



Australian Government

**Australian Institute of
Health and Welfare**

The Gap

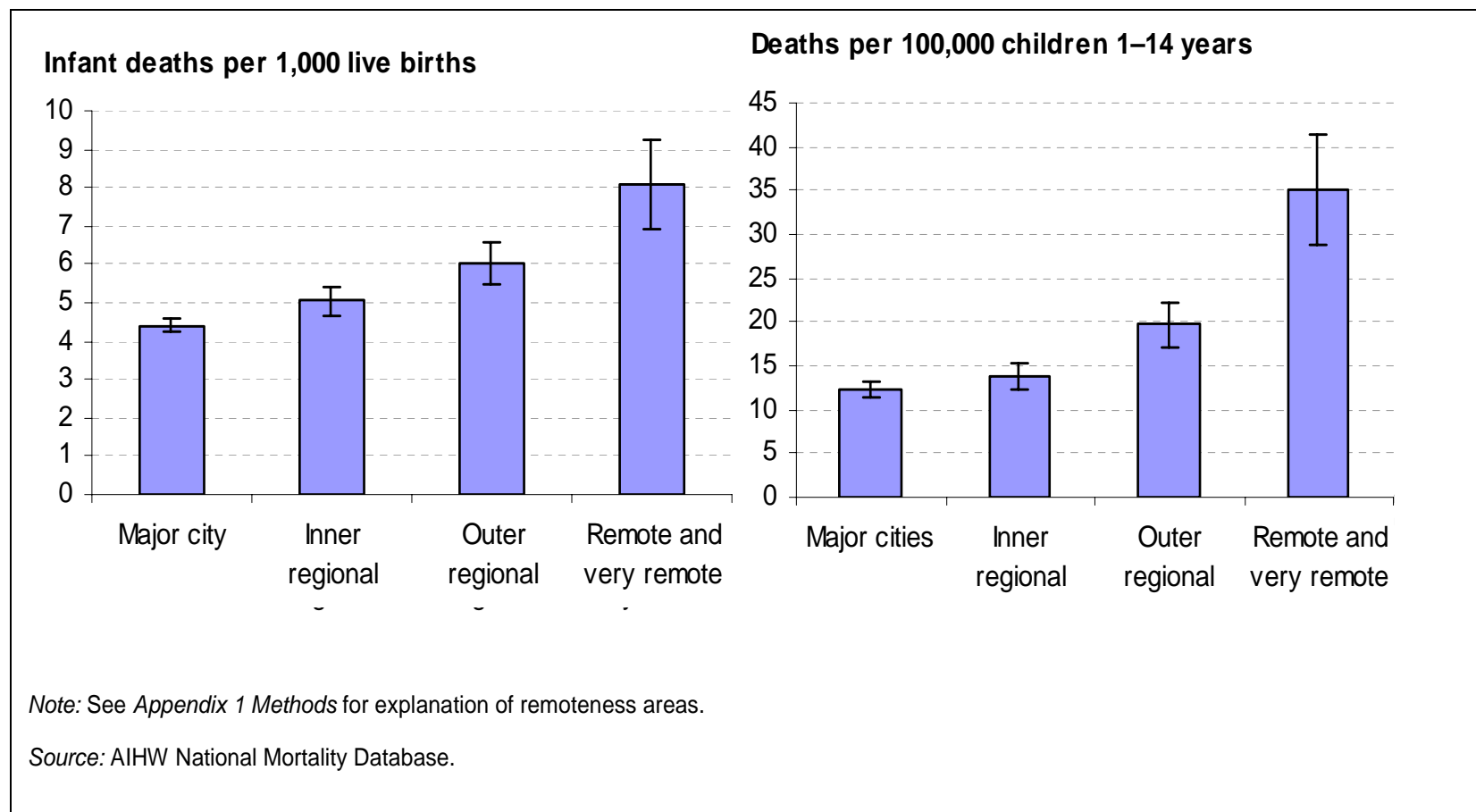


Figure 4.6: Infant and child deaths by remoteness, 2004–2006



Australian Government

**Australian Institute of
Health and Welfare**

9 m/o child Gardner's Syndrome/ APC mutation

Ref. Range Unit
<18 ug/L

CVa 5%
CVb 12%

| | | | | |
|------|-----|---------|-----|-----|
| Age: | 9 m | 8 1/2 m | 8 m | 6 m |
| AFP: | 137 | 132 | 85 | 53 |

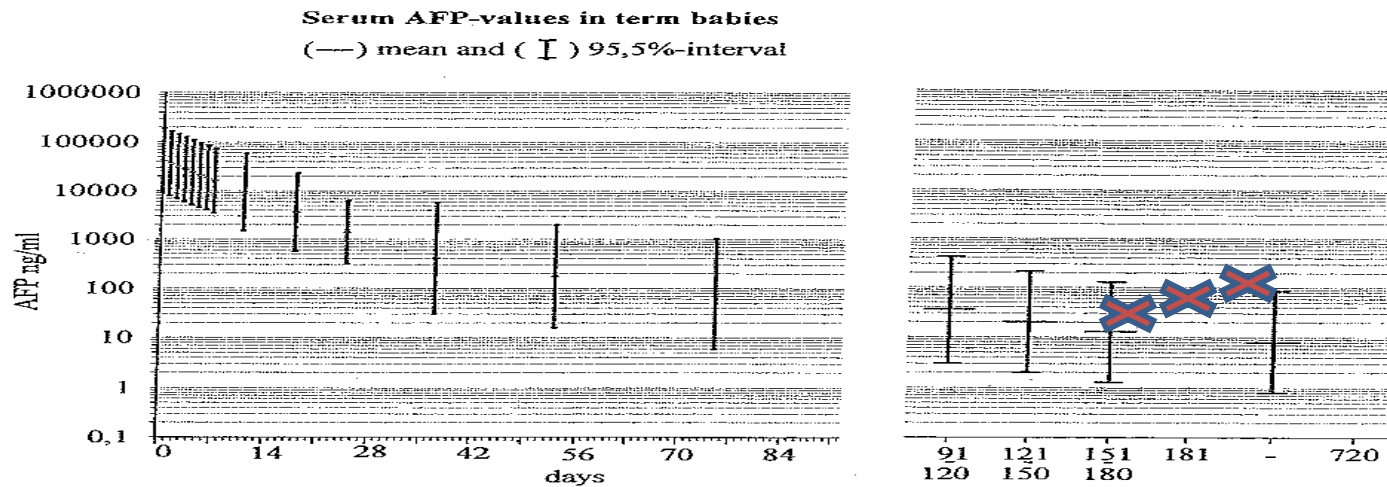


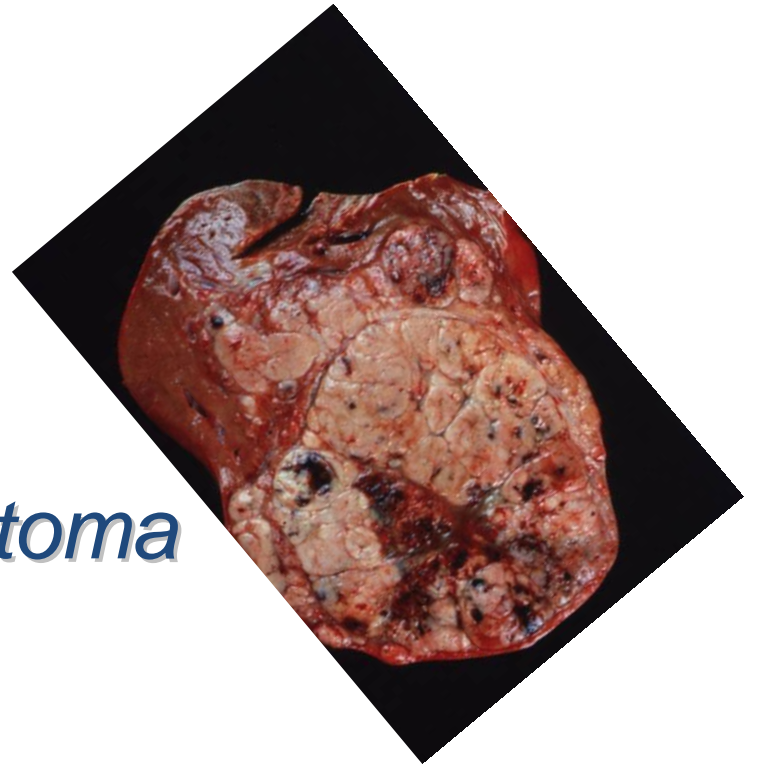
Figure 1. Serum AFP values in term babies: (—) mean and (I) 95.5% interval.

5/3/08 CT & MRI reveals:

15 mm hepatic lesion

19/03/08 Wedge resection:

*liver mass, hepatoblastoma
completely excised*

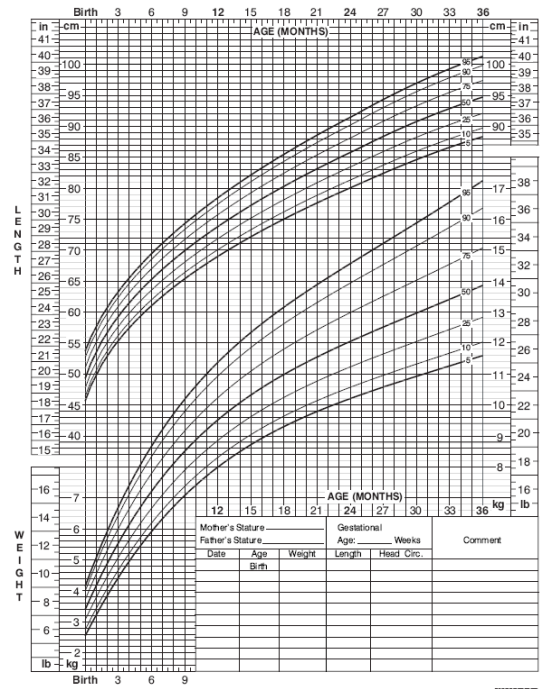


**DIAGNOSTIC VALUE OF ALPHA1-FETOPROTEIN
AND BETA-HUMAN CHORIONIC GONADOTROPIN IN
INFANCY AND CHILDHOOD**

Pediatric Hematology and Oncology, 2001; 18:11



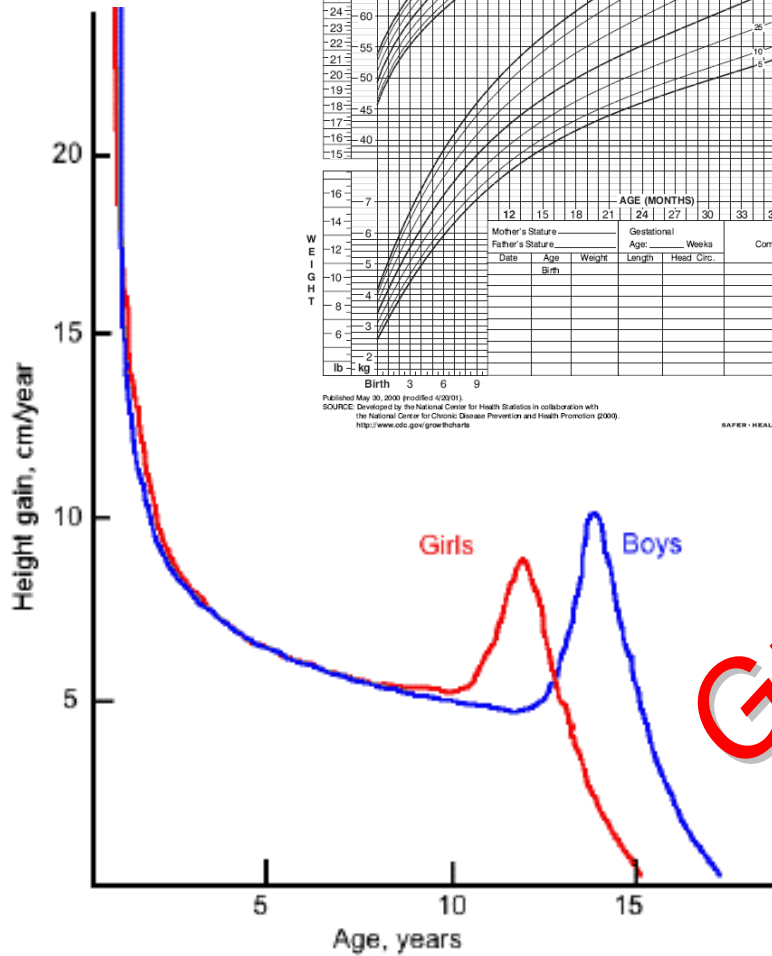
Birth to 36 months: Girls
Length-for-age and Weight-for-age percentiles



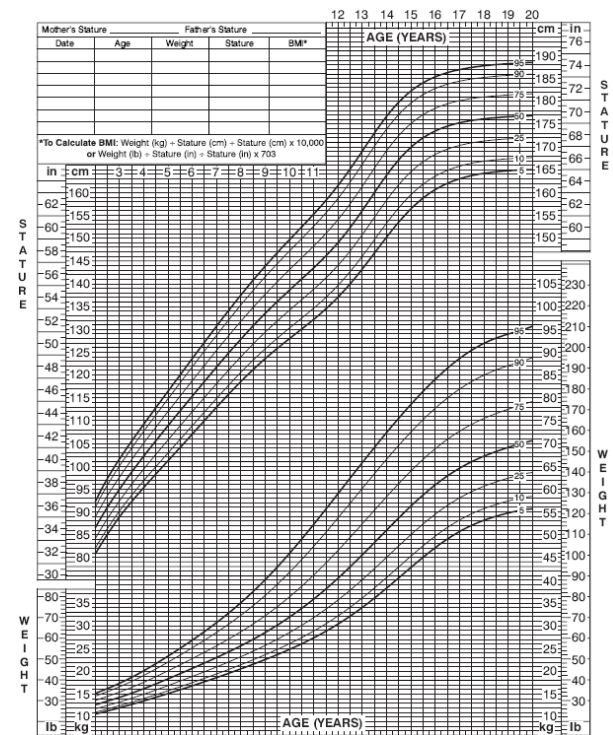
Published May 30, 2000 (modified 4/2001).
SOURCE: Developed by the National Center for Health Statistics in collaboration with the National Center for Chronic Disease Prevention and Health Promotion (2000).
<http://www.cdc.gov/growthcharts>

Growth

Tanner Stage



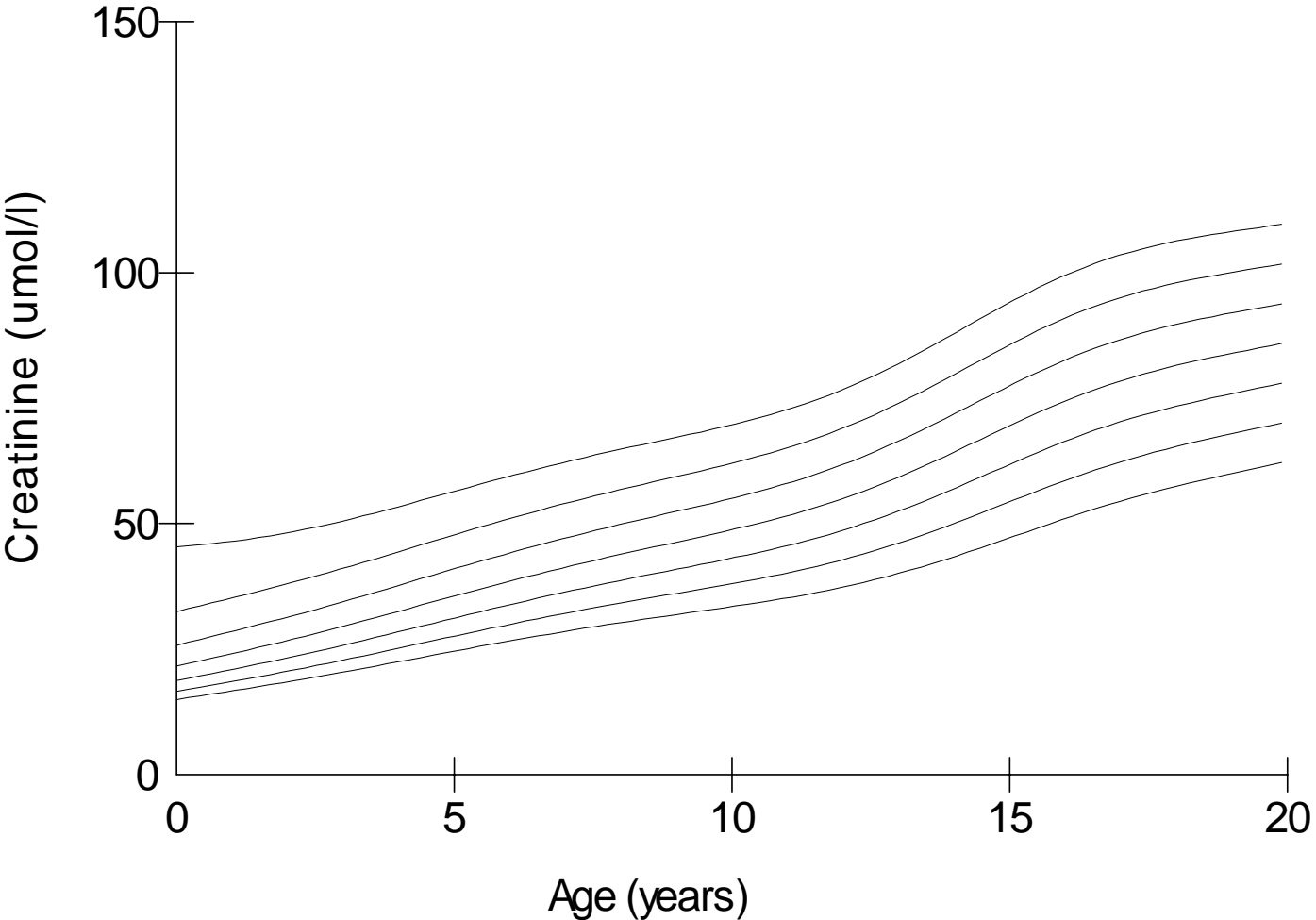
2 to 20 years: Boys
Stature-for-age and Weight-for-age percentiles

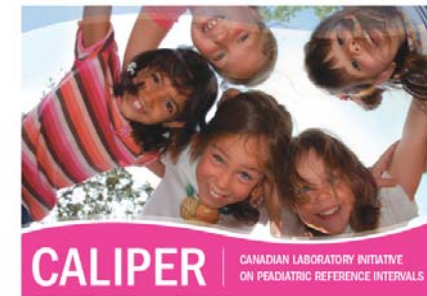
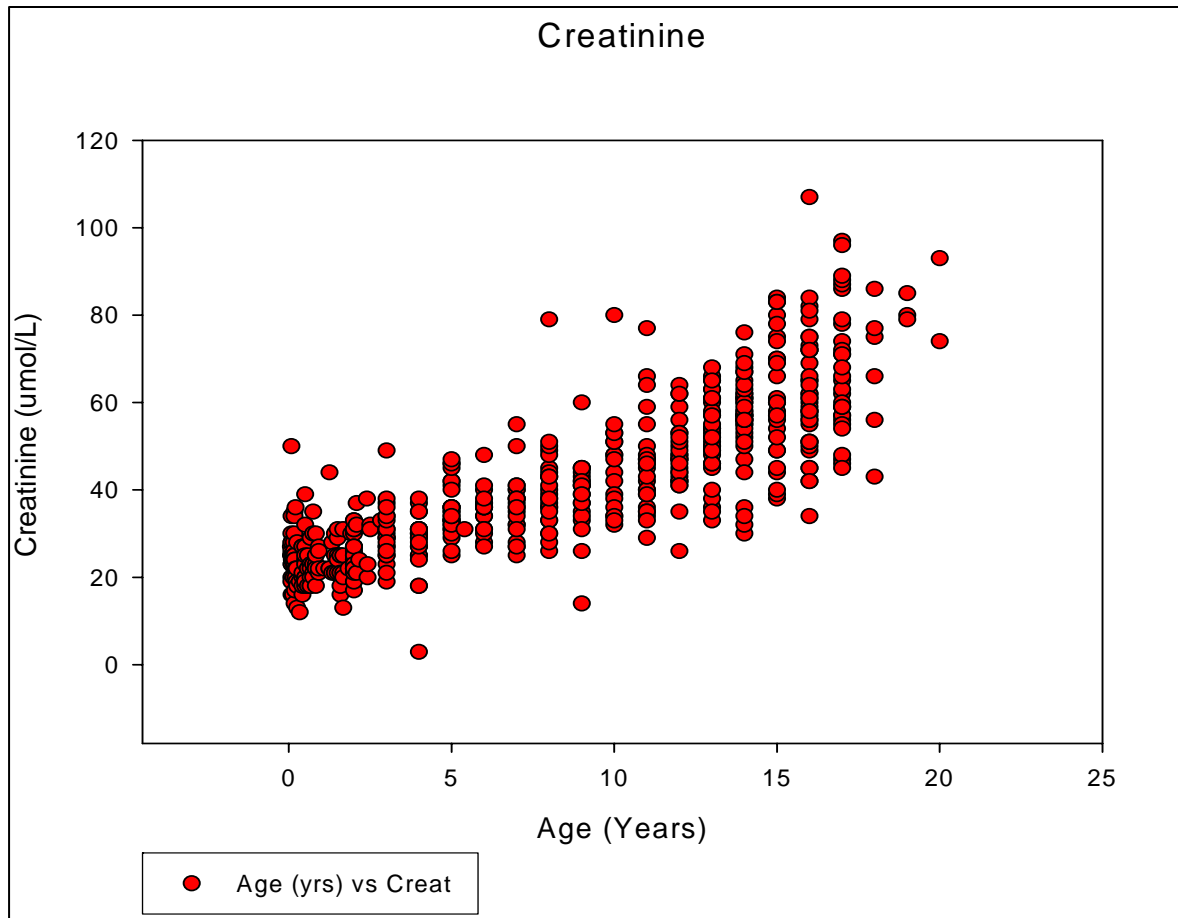


Published May 30, 2000 (modified 11/21/03).
SOURCE: Developed by the National Center for Health Statistics in collaboration with the National Center for Chronic Disease Prevention and Health Promotion (2000).
<http://www.cdc.gov/growthcharts>

SAFER · HEALTHIER · PEOPLE

Boys Creatinine





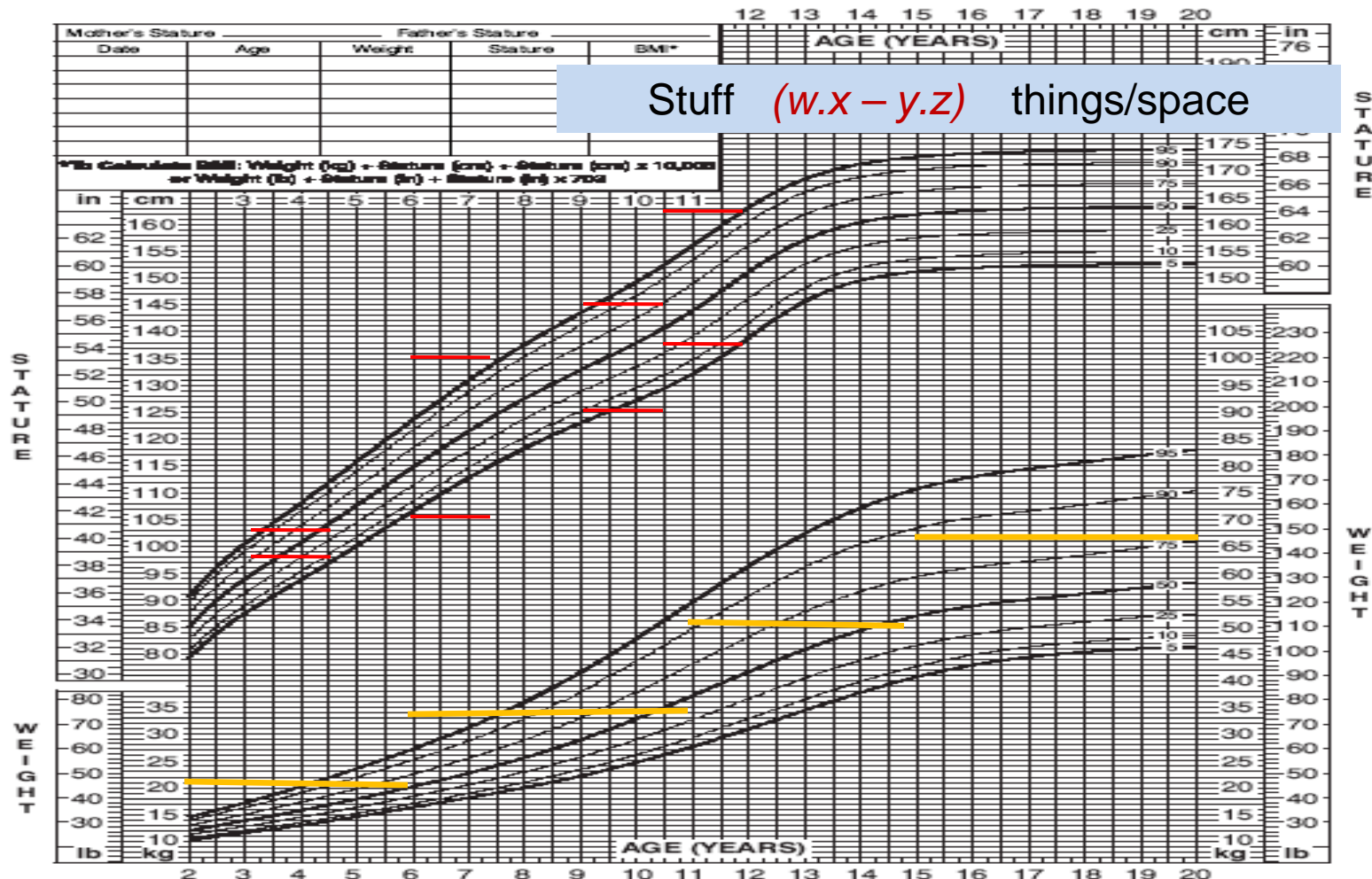
Pediatric Reference Intervals

Cobas 6000

| | Birth–12 mos | 1–5 years | 6-10 years | 11–14 years | 15–20 years |
|--|---------------------|------------------|-------------------|--------------------|--------------------|
| Creatinine (μmol/L) IDMS-Traceable | 14 – 39 (n=34) | 6 – 49 (n=46) | 25 – 60 (n=42) | 29 – 70 (n=71) | 42 – 100 (n=58) |

Discontinuous reference intervals for continuous variables.

2 to 20 years: Girls Stature-for-age and Weight-for-age percentiles NAME _____ RECORD # _____

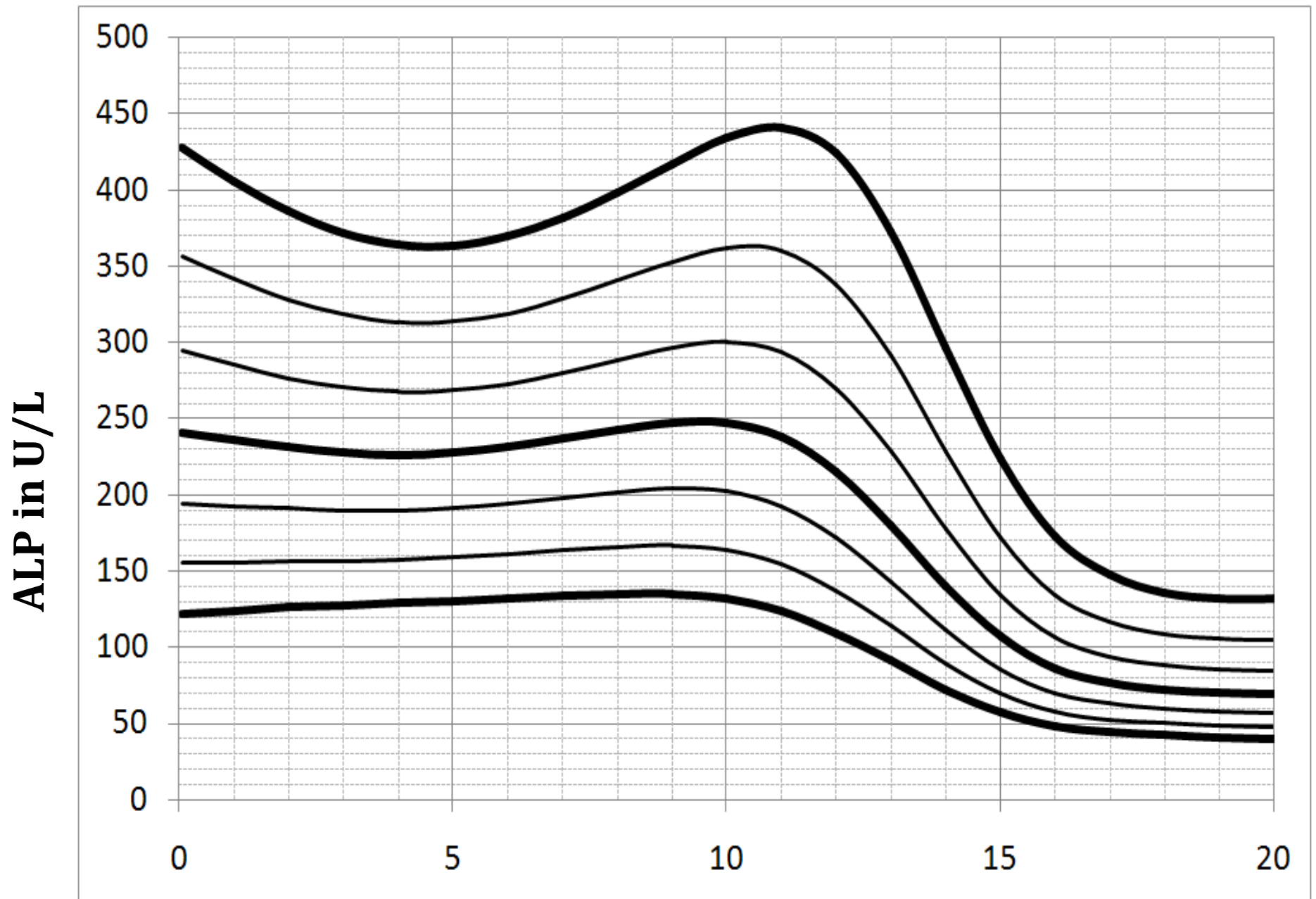


Published May 30, 2000 (modified 11/21/00).
SOURCE: Developed by the National Center for Health Statistics in collaboration with the National Center for Chronic Disease Prevention and Health Promotion (2000).
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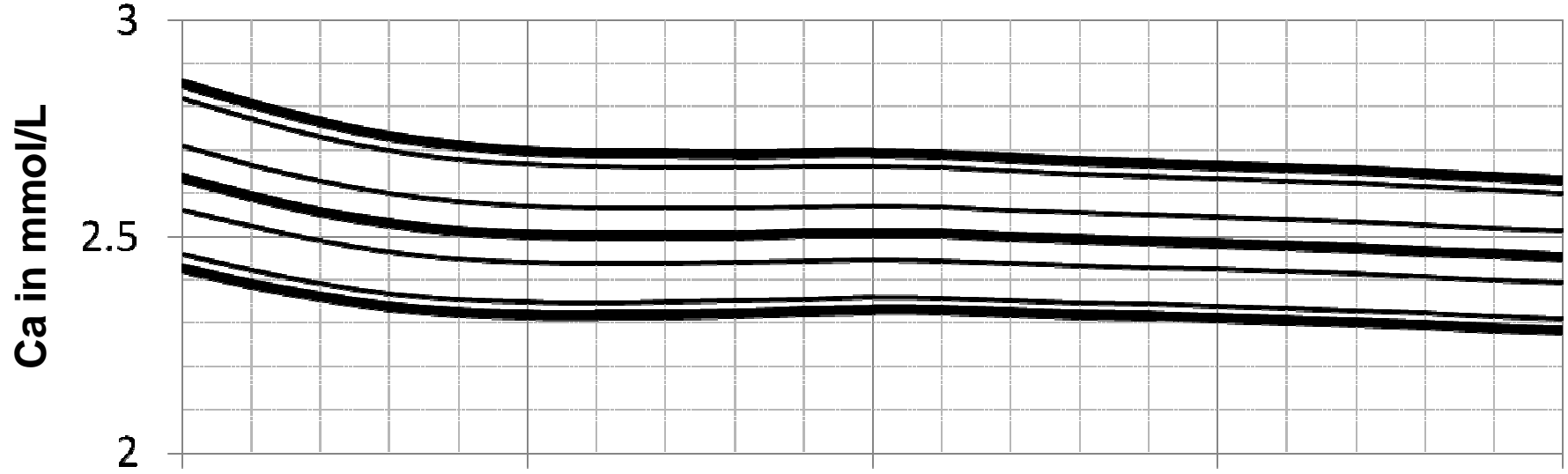


Partitioning.

| 9 year girl short | | Date: | 6/09/2009 | 8/07/2009 | 11/05/2009 | 6/05/2009 |
|-------------------|--------------------|---------------|---------------|---------------|--------------|---------------|
| | | Lab#: | 380456 | * 359605 | 340716 | * 825655 |
| | Ref. Range | Unit | | | | |
| Creat | (18-58) | umol/L | 21 | 29 | 30 | 24 |
| Urea | (2.3-7.5) | mmol/L | 3.6 | 3.6 | 4.4 | 3.3 |
| Calc Tot | (2.20-2.70) | mmol/L | 2.20 | 2.42 | 2.34 | 2.46 |
| Phosphate | (0.97-1.76) | mmol/L | L 0.76 | 1.14 | 1.05 | L 0.94 |
| Prot Tot | (63-83) | g/L | 65 | 81 | 79 | |
| Albumin | (35-50) | g/L | 43 | 49 | 47 | 47 |
| ALT | (5-45) | U/L | 10 | 18 | 17 | |
| GGT | (5-40) | U/L | 9 | 11 | 10 | |
| ALP | (95-390) | U/L | H 872 | H 1007 | H 788 | |
| Vitamin D | (50-150) | nmol/L | | | 85 | |
| PTH | (1.5-8.8) | pmol/L | | | H 15 | |



Girls' Alkaline Phosphatase



Girls' Phosphate & Calcium



| 9 year girl | short | Date: | 6/9/2009 | 8/7/2009 | 11/5/2009 | 6/5/2009 |
|------------------|--------------------|---------------|---------------|---------------|--------------|---------------|
| | | Lab#: | 380456 | * 359605 | 340716 | * 825655 |
| | Ref. Range | Unit | | | | |
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| Vitamin D | (50-150) | nmol/L | | | 85 | |
| PTH | (1.5-8.8) | pmol/L | | | H 15 | |

Autosomal dominant hypophosphataemic rickets

9 y/o boy

“Looks like the Michelin man”

PLASMA

| | | | |
|-------------|-----------|--------|-----|
| Creat | (18-58) | umol/L | 45 |
| Urea | (2.3-7.5) | mmol/L | 5.6 |
| Prot Tot | (63-83) | g/L | 41 |
| Albumin | (35-50) | g/L | 14 |
| Globulins | (22-38) | g/L | 27 |
| Cholesterol | (<4.5) | mmol/L | 12 |

URINE

| | | | |
|------------|-----------|---------|-------|
| Albumin | | mg/L | 2934 |
| Creatinine | | mmol/L | 4.0 |
| Alb/Cr | (0.2-2.9) | mg/mmol | 733.5 |
| Prot Tot | | g/L | 4.08 |
| Prot/Crea | (< 20) | mg/mmol | 1020 |

5 m/o boy
“looks starved”

| | | | |
|-----------|-------------|--------|------|
| Sodium | (135-145) | mmol/L | 124 |
| Potassium | (3.5-6.0) | mmol/L | 3.5 |
| Chloride | (97-109) | mmol/L | 88 |
| Bicarb | (15.0-29.0) | mmol/L | 27.5 |
| Creat | (18-58) | umol/L | 14 |
| Urea | (2.1-8.4) | mmol/L | 1.1 |
| Prot Tot | (50-78) | g/L | 40 |
| Albumin | (30-49) | g/L | 18 |
| Globulin | (17-29) | g/L | 22 |
| Chol | (< 4.50) | mmol/L | 7.15 |
| Trig | (< 1.70) | mmol/L | 7.30 |

URINE AND OTHER FLUIDS

| Spcm: | RandomUr | |
|------------------|----------|--------|
| Creat | mmol/L | 0.8 |
| Prot Tot | g/L | 18.25 |
| Prot/Crea (< 20) | mg/mmol | 22812 |
| Albumin | mg/L | > 4000 |
| Alb/Creat | mg/mmol | 8295 |

Rate of change of nephrotic proteinuria.

| | Date: | 24/09 | 18/08 |
|------------------|---------|--------|-------|
| Creat | mmol/L | 11 | 7.5 |
| Prot Tot | g/L | 16.2 | 0.49 |
| Prot/Crea (<20) | mg/mmol | 1473 | 65 |
| Albumin | mg/L | 15245 | 365 |
| Alb/Creat (<2.5) | mg/mmol | 1385.9 | 48.7 |

Proteinuria in Nephrosis

Heavy proteinuria (albuminuria) is the hallmark.
(Strictly speaking NO haematuria.)

Some children will excrete as much as
15 g/M²/24 hours.

The minimal protein excretion compatible is:

~ 1 g/M²/24 hours or

40 mg/M²/hour or

0.05g/kg body weight/24 hours

Nephrotic Syndrome/Proteinuria Criteria

Another criteria-
softer but functional.

Enough proteinuria to result in

1. Hypoalbuminaemia
2. Oedema
3. Hypercholesterolaemia
4. Hypercoagulability (AT3 deficiency)



Nephrosis/Nephrotic Syndrome

Incidence is 2-5 per 100,000 children < 16 years.

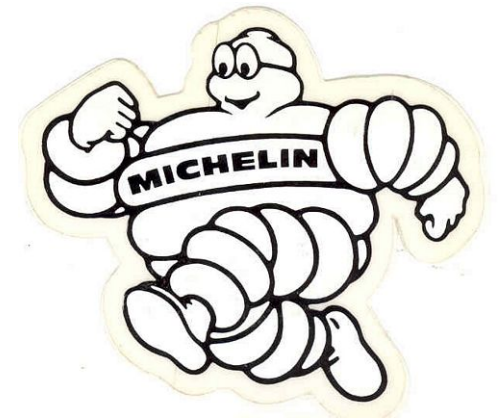
Prevalence is 15.5 per 100,000 individuals.

75% < 6 years with peak age of onset 2-3 years.

Earlier the onset, the greater the likelihood
the lesion is MCNS.

Onset <5 years, >90% MCNS.

Onset >10 years, MCNS ~50%,
FSGS ~30% and MPGN ~20%.



Nephrotic Syndrome

Mortality rate depends upon the disorder causing nephrosis.

With MCNS (minimal change nephrotic syndrome – nil lesion) long-term remission is greater than 70% with a cumulative mortality rate (at 20 y post onset) of less than 15% .

With FSGS (focal segmental glomerulosclerosis) 24% remission rate after 20 years with a cumulative mortality rate of greater than 50%.

24 year boy: RELAPSE OF NEPHROTIC SYNDROME

| | | |
|--------------------|-----------------------|-------------------|
| Urine Creatinine | 35.3 mmol/L | |
| Urine Protein | 91 g/L | (0-0.16) |
| Protein/Creatinine | 2600 (mg/mmol) | (<35) |
| Urine Albumin | 57 g/L | (<20.0) |
| Albumin/Creatinine | 1628 (mg/mmol) | (<3.5) |
| Urea | 10.5 mmol/L | (2.5-8.5) |
| Creatinine | 173 umol/L | (50-110) |
| Cholesterol | 10.3 mmol/L | (<5.5) |
| Tot Protein | 56 g/L | (65-85) |
| Albumin | 27 g/L | (36-46) |
| Globulins | 29 g/L | (25-36) |

Morbidity

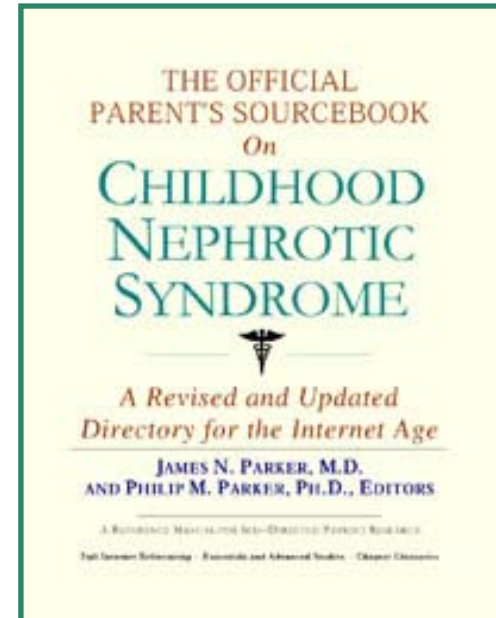
A prolonged period of treatment including frequent monitoring by parents and by doctor.

Administration of drugs associated with significant adverse events.

A high rate of recurrence (relapses in >60% of patients).

The potential for progression to chronic renal failure.

Clots (ATIII) & sepsis (complement).



6 y/o boy polyuria & polydipsia

| | | | |
|-----------|-------------|--------|------|
| Sodium | (135-145) | mmol/L | 137 |
| Potassium | (3.5-5.0) | mmol/L | 4.1 |
| Chloride | (97-109) | mmol/L | 101 |
| Bicarb | (15.0-29.0) | mmol/L | 27.8 |
| Anion Gap | (4.0-18.0) | | 12.3 |
| Creat | (18-58) | umol/L | 30 |
| Urea | (2.1-8.4) | mmol/L | 3.5 |
| Glucose | (3.0-11.0) | mmol/L | 20.4 |

12 y/o girl feeling poorly

| | | | |
|-----------|-------------|--------|-------------|
| Sodium | (135-145) | mmol/L | 141 |
| Potassium | (3.5-5.0) | mmol/L | 4.5 |
| Chloride | (97-109) | mmol/L | 106 |
| Bicarb | (19.0-30.0) | mmol/L | 10.8 |
| Anion Gap | (4.0-18.0) | | 28.7 |
| Creat | (31-77) | umol/L | 51 |
| Urea | (2.3-7.5) | mmol/L | 4.8 |

Increased anion gap acidosis.

12 y/o girl feeling poorly (2)

| | | | |
|-----------|-------------|--------|-------------|
| Lactate | (0.6-2.4) | mmol/L | 2.1 |
| Glucose | (<11.0) | mmol/L | 23.2 |
| GASES/pH | Spcm: VenBl | | |
| Base Exc | (-2-2) | mmol/L | -16 |
| pH | (7.34-7.45) | | 7.19 |
| pCO2 | (34-45) | mmHg | 31 |
| pO2 | (83-108) | mmHg | 34 |
| HCO3 Calc | (20.0-28.0) | mmol/L | 11.2 |

3 y/o boy Polyuria & Polydipsia

| | | | |
|-----------------------|-------------|--------|--------------|
| Sodium | (135-145) | mmol/L | 132 |
| Potassium | (3.5-5.0) | mmol/L | 4.9 |
| Chloride | (97-109) | mmol/L | 100 |
| Bicarb | (15.0-29.0) | mmol/L | (cap sample) |
| Creat | (18-58) | umol/L | 13 |
| Urea | (2.1-8.4) | mmol/L | 6.7 |
| Glucose | (2.5-11.0) | mmol/L | 31.6 |
| pH | (7.34-7.45) | | 7.40 |
| HCO ₃ Calc | (20.0-28.0) | mmol/L | 24.9 |

19 y/o girl known DM

| | | | |
|-----------|-------------|--------|-------------|
| Sodium | (135-145) | mmol/L | 137 |
| Potassium | (3.5-5.0) | mmol/L | 4.6 |
| Chloride | (97-109) | mmol/L | 100 |
| Bicarb | (18.0-31.0) | mmol/L | 8.2 |
| Anion Gap | (4.0-18.0) | | 33.4 |
| Creat | (44-100) | umol/L | 70 |
| Urea | (2.7-7.2) | mmol/L | 5.4 |

19 y/o girl known (2)

| | | | |
|---------|-----------|--------|-------------|
| Lactate | (0.6-2.4) | mmol/L | 1.8 |
| Glucose | (<11.0) | mmol/L | 20.4 |

| | | | |
|-----------------------|-------------|-------------|-------------|
| GASES/pH | | Spcm: capBI | |
| Base Exc | (-2-2) | mmol/L | -22 |
| pH | (7.34-7.45) | | 7.03 |
| pCO ₂ | (34-45) | mmHg | 36 |
| HCO ₃ Calc | (20.0-28.0) | mmol/L | 11.2 |

8 y/o “fasting”

| Time | | | 945 | 1930 |
|-------------|------------|--------|-------------|-------------|
| Sodium | (135-145) | mmol/L | 138 | 126 |
| Potassium | (3.5-5.0) | mmol/L | 4.9 | 4.7 |
| Chloride | (97-109) | mmol/L | 90 | 85 |
| Bicarb | (18.-31.) | mmol/L | 22 | 16 |
| Anion Gap | (4.0-18.0) | | 30.9 | 30 |
| Glucose | (3.2-11.0) | mmol/L | 28.5 | 50 |
| AcAc | (<0.1) | mmol/L | | 2.6 |
| BHB | (<0.3) | mmol/L | | 7.1 |

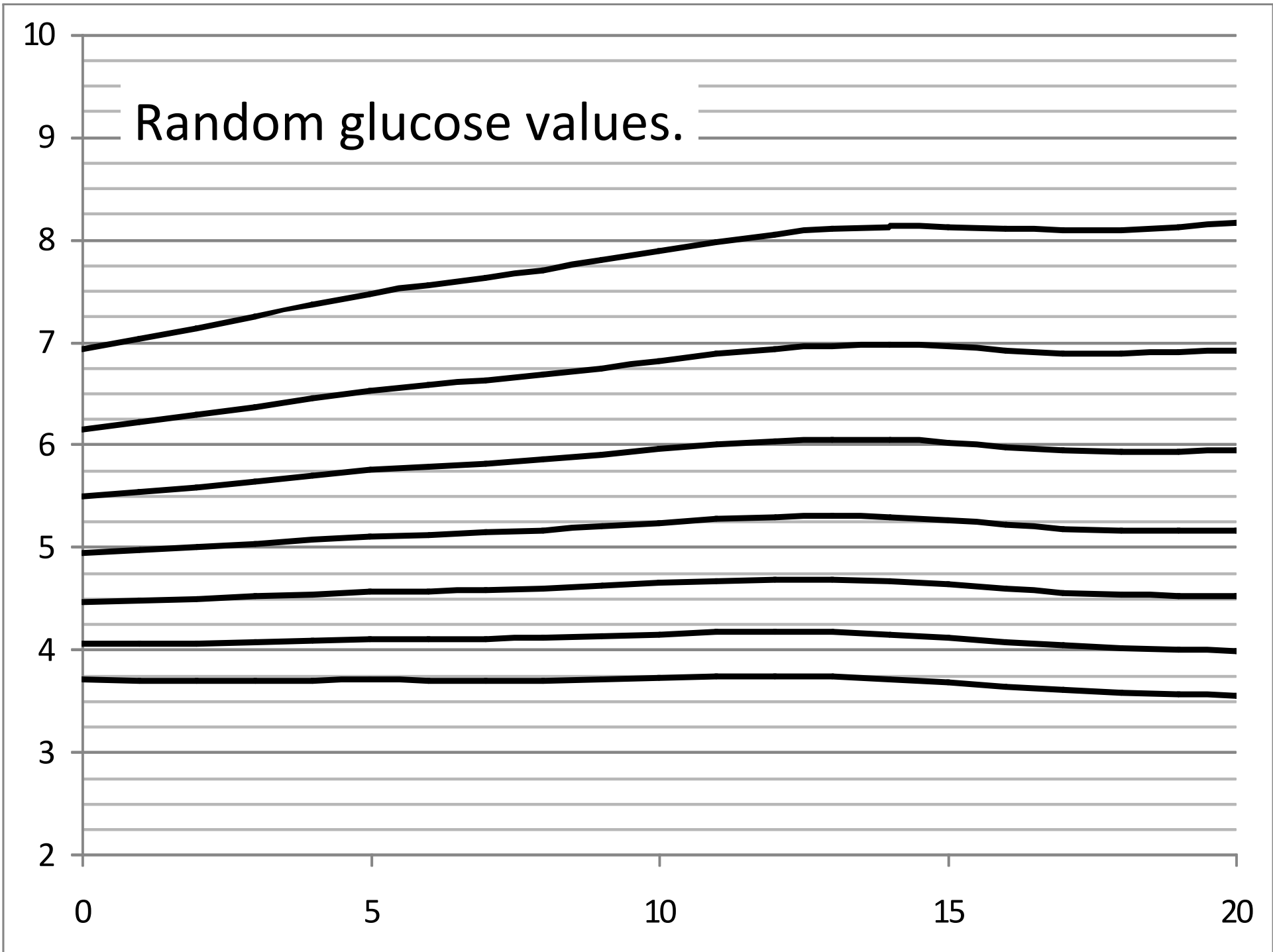
Diagnostic Criteria DKA

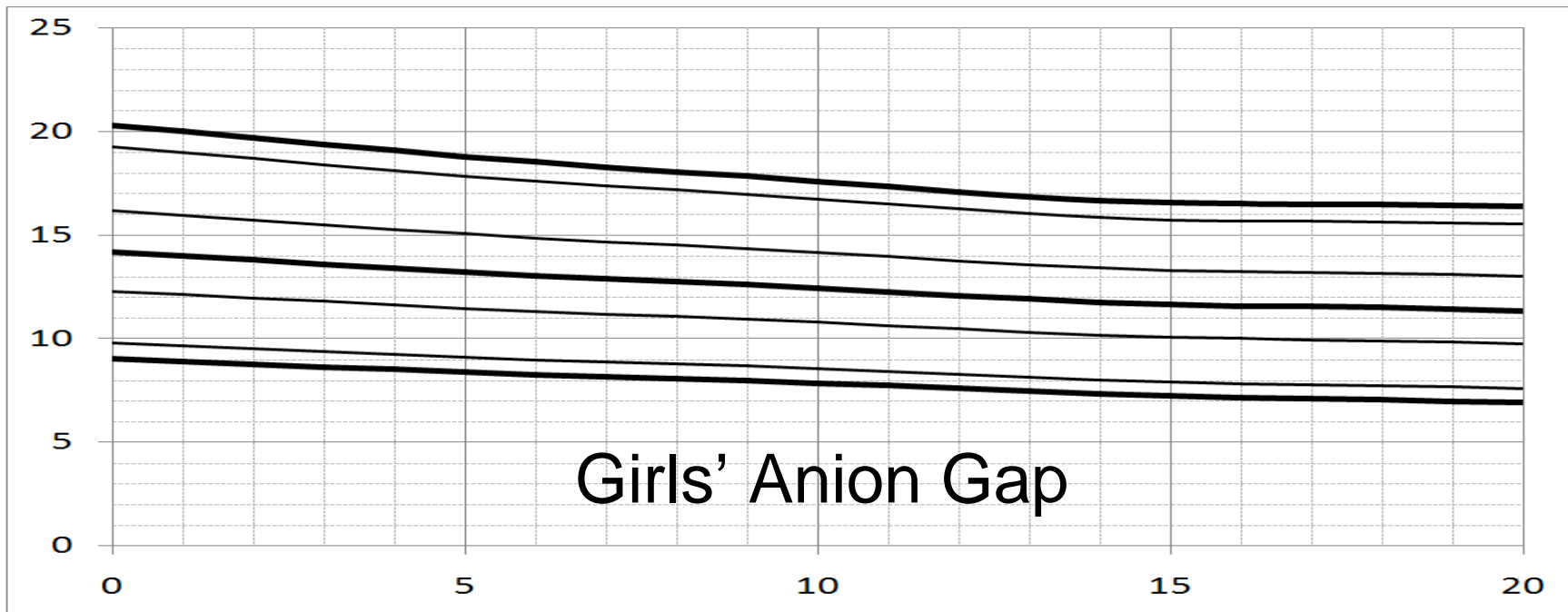
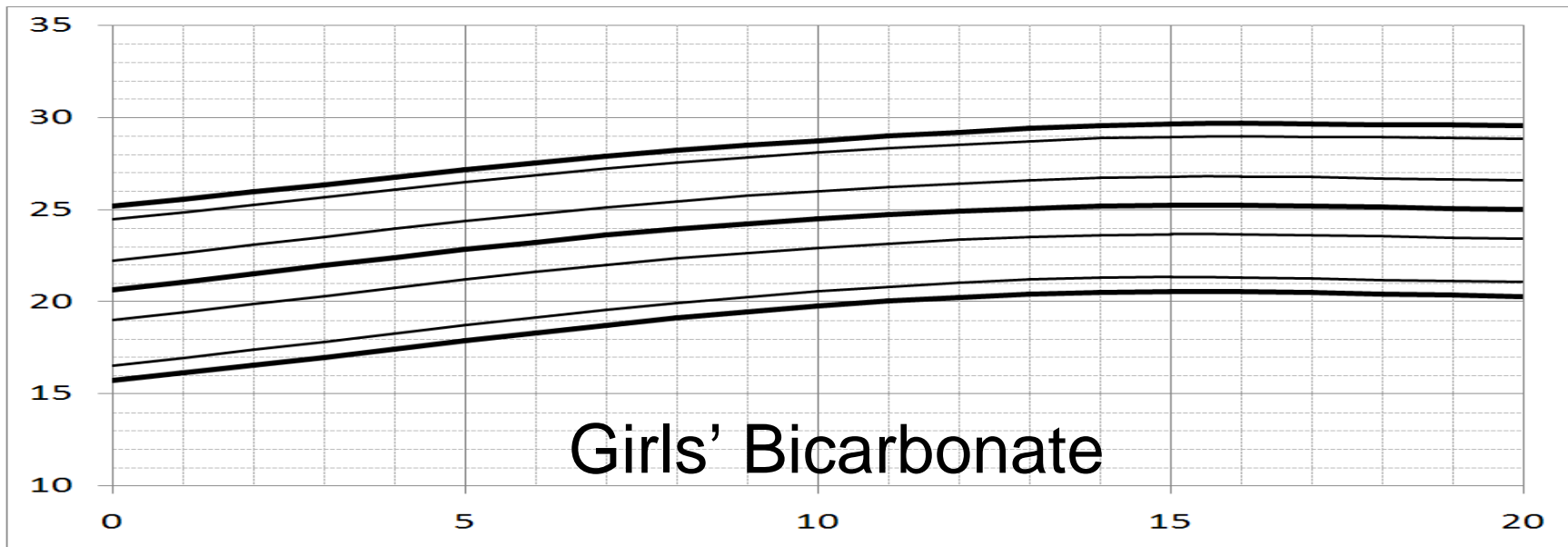
Glucose > 14mmol/l

$\text{HCO}_3^- < 15\text{mmol/l}$

pH < 7.25

Increased anion gap.





4 y/o boy V&D KA

| | | | |
|------------------|-------------|--------|-------------|
| Sodium | (135-145) | mmol/L | 133 |
| Potassium | (3.5-5.0) | mmol/L | 4.8 |
| Chloride | (97-109) | mmol/L | 95 |
| Bicarb | (15.0-29.0) | mmol/L | 13.4 |
| Anion Gap | (4.0-18.0) | | 29.4 |
| pH | (7.34-7.45) | | 7.37 |
| pCO ₂ | (34-45) | mmHg | 28 |
| Glucose | (3.0-11.0) | mmol/L | 4.3 |

3 y/o girl V&D KA

| | | | |
|-----------|-------------|--------|-------------|
| Sodium | (135-145) | mmol/L | 138 |
| Potassium | (3.5-5.0) | mmol/L | 4.6 |
| Chloride | (97-109) | mmol/L | 102 |
| Bicarb | (15.0-29.0) | mmol/L | 6.5 |
| Anion Gap | (4.0-18.0) | | 34.1 |
| Creat | (18-58) | umol/L | 57 |
| Urea | (2.1-8.4) | mmol/L | 13.1 |
| Glucose | (3.0-11.0) | mmol/L | 3.5 |

Type 1 Diabetes Mellitus

Very unpredictable.

Ketoacidosis with (relatively) low glucose.

Marked hyperglycaemia with no ketoacidosis.

Polyuria & polydipsia. (Confused with DI)

Weight loss.

Fatigue, malaise.

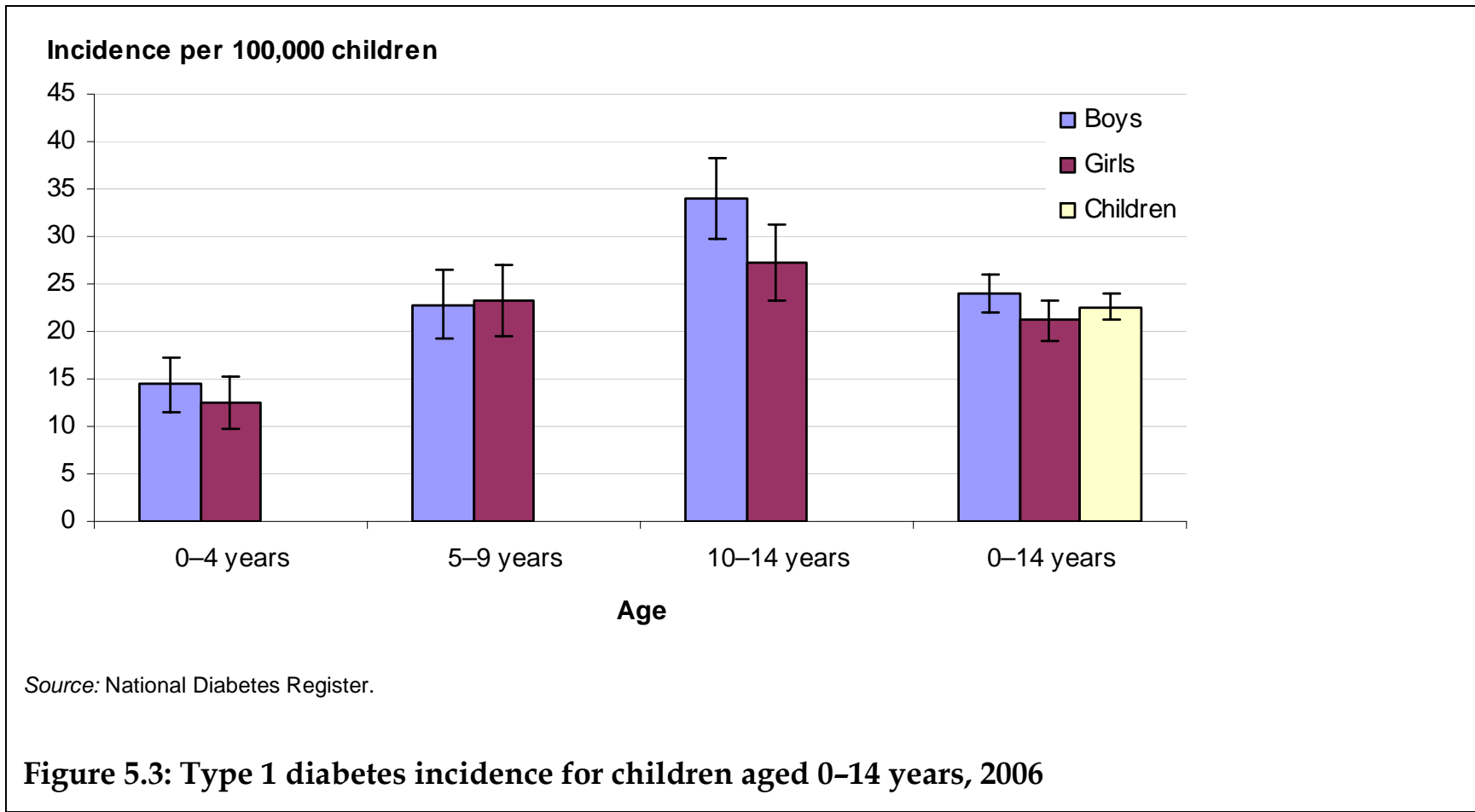
Type 1 Diabetes Mellitus

Diabetic Ketoacidosis

Incidence ~2 episodes per 100 patient years of diabetes. Up to 30% of presentations.

Mortality rate of DKA is about 2% per episode.

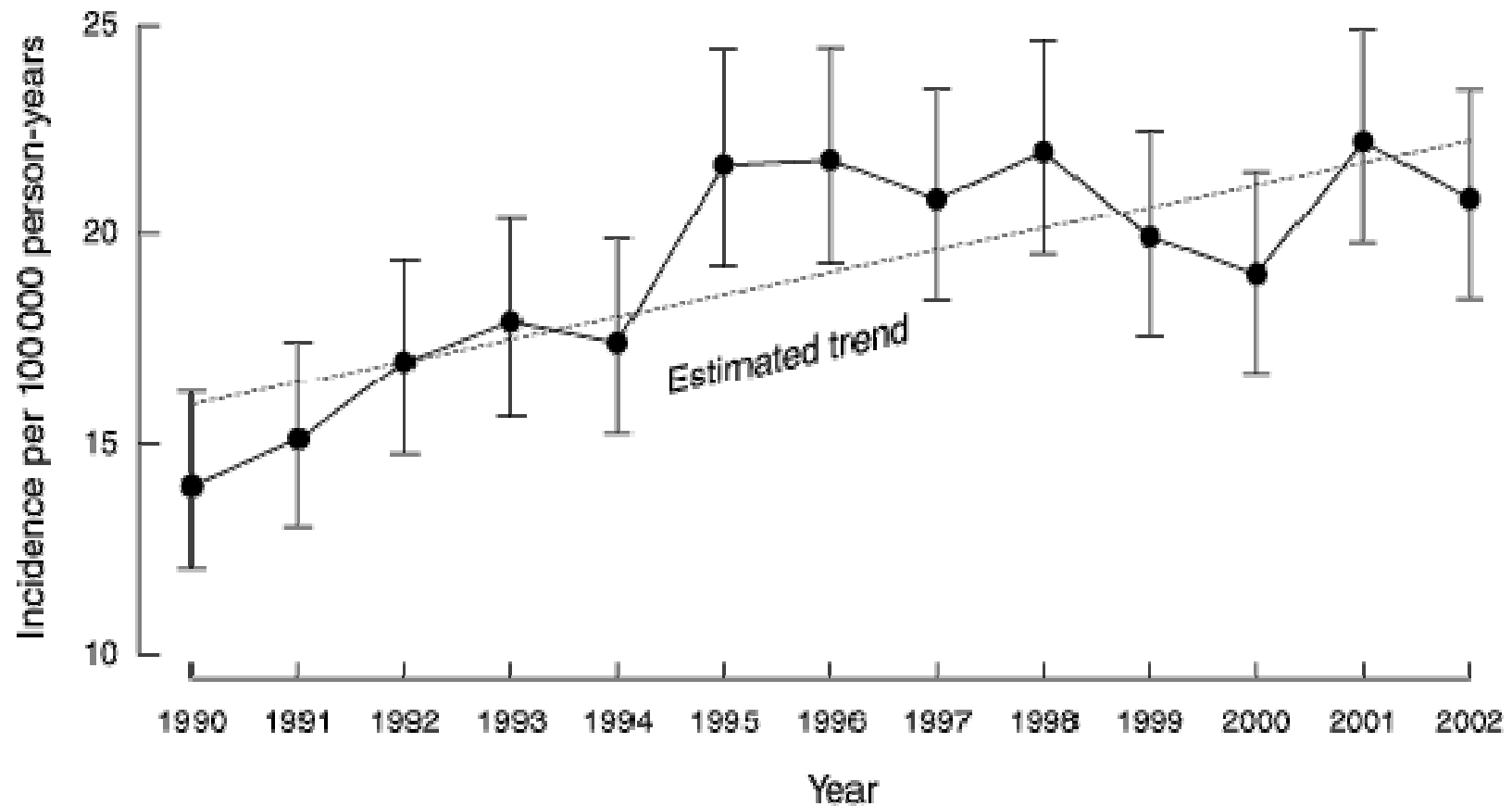
Death in DKA is mostly due to cerebral edema.



In South Australia, about 1 per week.



Increase in incidence of Type 1 DM



MJA 2005; 183 (5): 243-246

Oral glucose tolerance test

5 year girl 15 kg

Clin Notes: POLYURIA DEC WT DIZZINESS
?ASCITES

Test Req: OGTT, E/LFT, CBP

All but no role for OGTT in children

17 YEAR BOY

Clin notes: POLYURIA POLYDYPسيا

| Time | Glucose (mmol/L) |
|---------|---------------------|
| Fasting | 4.6 |
| 2 hr | 4.7 |

All but no role for OGTT in children

7 year boy: BSL 31.2 DM-2

| | | | |
|---------------------|------|--------|------------|
| Glucose | 10.7 | mmol/L | (3.6-6.0) |
| Glucose | 32.2 | mmol/L | (3.6-6.0) |
| β OH butyrate | 1.2 | mmol/L | (<0.3) |
| Acetoacetate | 0.5 | mmol/L | (<0.1) |
| C-Peptide | 141 | pmol/L | (300-1600) |
| Insulin | 5 | uU/ml | (1-14) |

All but no role for OGTT in children

2 y/o boy pale and unsettled

| | | | | |
|-------|---------------|-----------------|--------|-------------|
| LDH | (70-260) | | U/L | 1279 |
| Urate | (0.12-0.32) | | mmol/L | 0.27 |
| HGB | (105-125) | | g/L | 74 |
| PLTS | (150-450) | $\times 10^9/L$ | | 11 |
| WBC | (6.00-12.00) | $\times 10^9/L$ | | 85.5 |
| Blast | (0.00 - 0.01) | $\times 10^9/L$ | | 66.69 |

5 year boy pale

| | | | |
|-------|---------------|-----------------|-------------|
| LDH | (70-260) | U/L | 731 |
| Urate | (0.12-0.32) | mmol/L | 0.33 |
| HGB | (116-136) | g/L | 60 |
| PLTS | (150-450) | $\times 10^9/L$ | 18 |
| WBC | (6.10-11.00) | $\times 10^9/L$ | 402 |
| Blast | (0.00 - 0.01) | $\times 10^9/L$ | 389.94 |

9 year boy gait disturbance

| | | | | |
|----------|-------------|--------|---|-------------|
| Urate | (0.12-0.32) | mmol/L | | 0.32 |
| Prot Tot | (50-78) | g/L | | 74 |
| Albumin | (33-49) | g/L | | 42 |
| AST | (5-40) | U/L | H | 74 |
| ALT | (5-45) | U/L | | 12 |
| GGT | (5-40) | U/L | | 10 |
| LDH | (70-260) | U/L | H | 2155 |
| ALP | (95-390) | U/L | | 186 |
| CK | (25-200) | U/L | | 146 |

Childhood Cancer

Among the 12 major types of childhood cancers, leukemias and brain tumours account for more than half of the new cases.

About one-third of childhood cancers are leukemias. The most common type of leukemia in children is acute lymphoblastic leukemia.

The most common solid tumors are brain tumors (e.g., gliomas and medulloblastomas), with other solid tumors (e.g., neuroblastomas, Wilms tumors, and sarcomas such as rhabdomyosarcoma and osteosarcoma) being less common.

Cancer

Table 3: Number of new cancer cases for children aged 0–14, selected cancers and total cancers, by sex and age group, 2001

| Type of cancer | Males | | | Females | | | Total |
|--|-------|-----|-------|---------|-----|-------|-------|
| | 0–4 | 5–9 | 10–14 | 0–4 | 5–9 | 10–14 | 0–14 |
| Leukaemia | 63 | 33 | 26 | 47 | 30 | 26 | 225 |
| Brain and other central nervous system | 25 | 28 | 15 | 27 | 13 | 8 | 116 |
| All lymphomas | 9 | 13 | 24 | 3 | 8 | 11 | 68 |
| Connective and other soft tissue | 13 | 3 | 2 | 11 | 1 | 7 | 37 |
| Kidney, ureter and urethra | 11 | 1 | 0 | 15 | 4 | 1 | 32 |
| Other endocrine glands | 10 | 3 | 0 | 7 | 1 | 0 | 21 |
| Bone and articular cartilage | 0 | 5 | 2 | 2 | 4 | 8 | 21 |
| Eye | 9 | 1 | 0 | 7 | 1 | 2 | 20 |
| Skin-melanoma | 0 | 0 | 6 | 1 | 6 | 6 | 19 |
| All cancers (except NMSC) | 156 | 89 | 79 | 128 | 72 | 79 | 603 |

Note: NMSC Non-melanocytic skin cancer.

~ 3,300,000 children

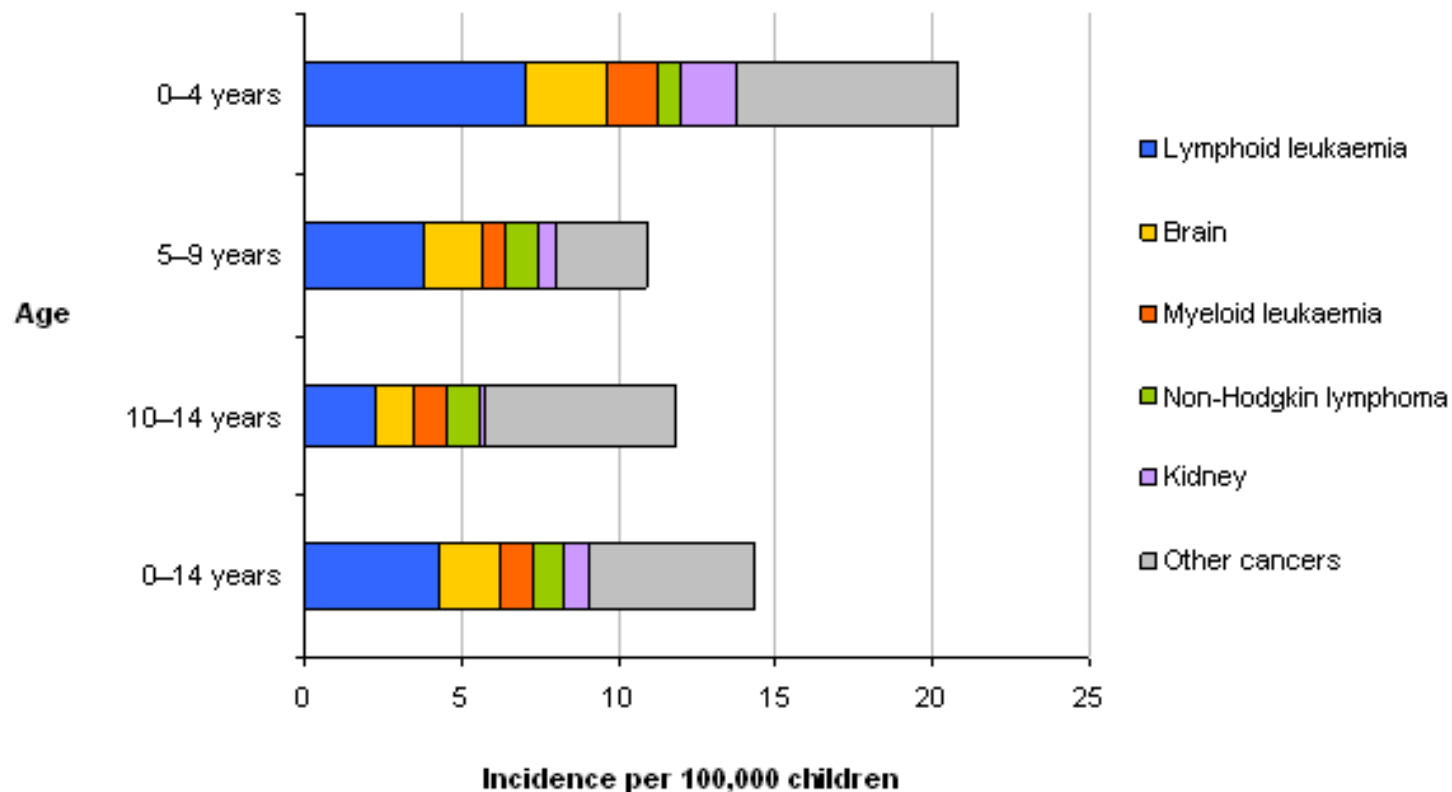
Leukaemia & Lymphoma ~1/10k, Brain tumours ~1/30k



Table 5.1: Cancer deaths among children aged 0–14 years, 1997–2006

| | 1997 | 1998 | 1999 | 2000 | 2001 | 2002 | 2003 | 2004 | 2005 | 2006 |
|-----------------------------|------|------|------|------|------|------|------|------|------|------|
| Number | 140 | 154 | 122 | 106 | 118 | 124 | 102 | 114 | 96 | 90 |
| Deaths per 100,000 children | 3.6 | 3.9 | 3.1 | 2.7 | 3.0 | 3.1 | 2.5 | 2.8 | 2.4 | 2.2 |

Note: Refer to Table A1.2 for ICD-9 and ICD-10 codes.



Most common types of new cancers among children aged 0–14 years, 2001–2005



Australian Government

Australian Institute of Health and Welfare

Cancer

- Tumour lysis
- Uricase
- High dose methotrexate
- Chemotherapy side effects
- Treatment late effects

10 year old girl Rhabdomyolysis

| | | | | |
|-------------|----------|--------|-------|-------|
| Tot Protein | (63-83) | g/L | 52 | 54 |
| Albumin | (35-50) | g/L | 30 | 30 |
| Bili Tot | (2-20) | umol/L | 24 | 20 |
| ALT | (5-45) | U/L | 570 | 635 |
| GGT | (5-40) | U/L | 15 | 15 |
| ALP | (95-390) | U/L | 78 | 94 |
| CK | (25-200) | U/L | 65531 | 49679 |

Query mitochondrial disease

10 year boy

Date: 03/09/08 02/09/08 01/09/08

CK (25-200) U/L

H 1431 H 4484 H 10396

viral myositis

4 y/o boy Weak

Date: 03/06/04 31/05/04

| | Ref. Range | Unit | |
|-----------------|------------------|---------------|------------------------|
| Creat | (18-58) | umol/L | 22 |
| Urea | (2.1-8.4) | mmol/L | 2.4 |
| Prot Tot | (63-83) | g/L | 69 |
| Albumin | (35-50) | g/L | 41 |
| Globulin | (20-36) | g/L | 28 |
| Bili Tot | (2-20) | umol/L | 8 |
| ALT | (5-45) | U/L | H 353 |
| GGT | (5-40) | U/L | 6 |
| CK | (25-200) | U/L | H 39355 H 15569 |

04-344607

4 y/o boy Gower's Sign

| | | | | |
|----------|-----------|--------|---|-------|
| Creat | (18-58) | umol/L | L | 12 |
| Urea | (2.1-8.4) | mmol/L | | 4.0 |
| Prot Tot | (50-78) | g/L | | 67 |
| Albumin | (33-49) | g/L | | 44 |
| Globulin | (20-36) | g/L | | 23 |
| Bili Tot | (< 20) | umol/L | | 3 |
| ALT | (5-45) | U/L | H | 713 |
| GGT | (5-40) | U/L | | 8 |
| ALP | (95-390) | U/L | | 115 |
| CK | (25-200) | U/L | H | 23193 |

16 year young man

Respiratory complaint

| | | | |
|----------|-----------|--------|----------------|
| Creat | (31-77) | umol/L | < 10 |
| Urea | (2.1-8.4) | mmol/L | 3.5 |
| TotProt | (63-83) | g/L | 87 |
| Albumin | (35-50) | g/L | 52 |
| Globulin | (22-34) | g/L | 35 |

Muscular Dystrophy



MUSCULAR DYSTROPHIES:

Duchenne Muscular Dystrophy (DMD)

(Also known as Pseudohypertrophic)

Becker Muscular Dystrophy (BMD)

Emery-Dreifuss Muscular Dystrophy (EDMD)

Limb-Girdle Muscular Dystrophy (LGMD)

Myotonic Dystrophy (MMD)

(Also known as Steinert Disease)

Oculopharyngeal Muscular Dystrophy (OPMD)

Distal Muscular Dystrophy (DD) (Miyoshi)

Congenital Muscular Dystrophy (CMD)

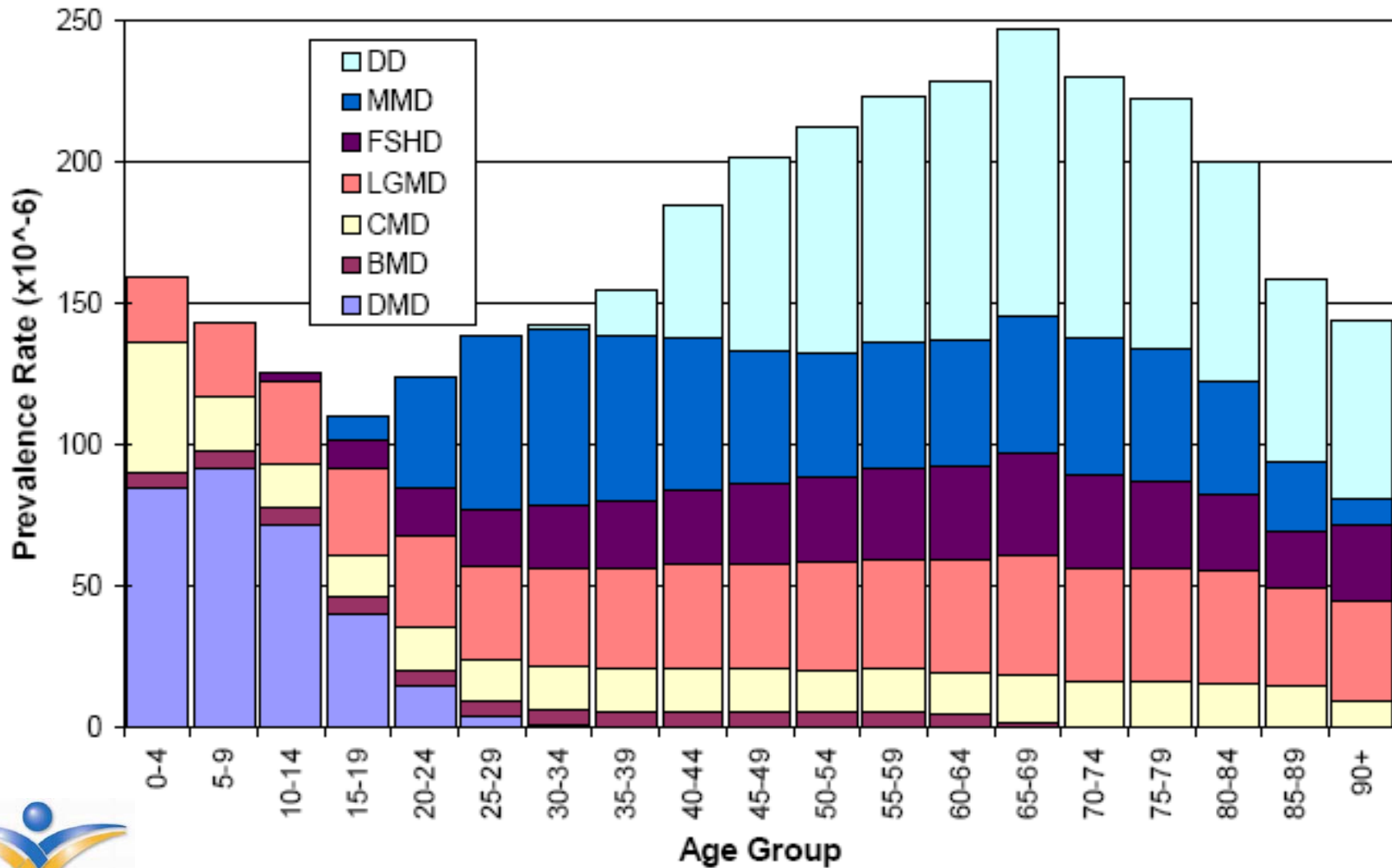
Facioscapulohumeral Muscular Dystrophy

(FSH or FSHD)(Also known as Landouzy-Dejerine)

Gower's Sign



PREVALENCE RATE OF ALL MD TYPES, 2005 (RATE PER MILLION)



MUSCULAR
DYSTROPHY
mda.org.au

Prepared by Access Economics for MDA
October 2007

Muscular Dystrophy

What about urine assays
reported as X / mmol creatinine?

Neonatal Hyperbilirubinaemia

“He who knows syphilis, knows medicine.”

Sir William Osler

“He who knows bilirubin, knows babies.”

Anonymous

Extensive differential diagnosis for *both direct and indirect* hyperbilirubinaemia.

5 week girl persistent jaundice

Happy, healthy, and robust.

| | | | | |
|-----------|----------|--------|---|-----|
| Prot Tot | (50-78) | g/L | | 58 |
| Albumin | (30-49) | g/L | | 35 |
| Globulin | (17-29) | g/L | | 23 |
| Bili Tot | (2-20) | umol/L | H | 200 |
| Bili Conj | (2-4) | umol/L | H | 183 |
| ALT | (5-80) | U/L | | 70 |
| GGT | (10-150) | U/L | H | 521 |
| ALP | (75-435) | U/L | H | 488 |

5 week girl persistent jaundice

Happy, healthy, and robust.

| | | | | |
|-----------|----------|--------|---|------|
| Prot Tot | (50-78) | g/L | | 58 |
| Albumin | (30-49) | g/L | | 36 |
| Globulin | (17-29) | g/L | | 22 |
| Bili Tot | (2-20) | umol/L | H | 160 |
| Bili Conj | (2-4) | umol/L | H | 94 |
| ALT | (5-80) | U/L | H | 105 |
| GGT | (10-150) | U/L | H | 1033 |
| ALP | (75-435) | U/L | H | 556 |

Direct bili should be < 20% of total bili.

Extrahepatic Biliary Atresia

- Extrahepatic Biliary Atresia (EBA) is a disorder characterised by atresia of the extrahepatic bile ducts.
- It is the most frequent surgical cause of cholestatic jaundice in this age group.
- 1/18,000 live births in Western Europe and higher in Asia and the Pacific region.
- Females are affected more often than males.

Current prognosis of BA

UK 1999–2002 (148 patients)

4-year survival with native liver

after Kasai Operation 51%

4-year survival

after liver transplantation 90%

Kasai works better before 8 weeks.

Children present with intracranial haemorrhage at 3-6 months due to Vitamin K malabsorption and deficiency.

Biliary Atresia Screening: Why, When, and How?

Sokol Pediatrics.2009; 123: 951-2.

10 day girl jaundice

| | | | | |
|-----------|----------|--------|---|-----|
| Prot Tot | (50-78) | g/L | L | 48 |
| Albumin | (30-49) | g/L | L | 26 |
| Globulin | (17-29) | g/L | | 22 |
| Bili Tot | (50-250) | umol/L | | 177 |
| Bili Conj | (2-4) | umol/L | H | 104 |
| ALT | (5-80) | U/L | H | 96 |
| GGT | (10-150) | U/L | | 93 |
| ALP | (75-435) | U/L | H | 436 |

α 1 antitrypsin deficiency

3 week boy

| | | | |
|-----------|----------|--------|-------------|
| Prot Tot | (50-78) | g/L | 36 |
| Albumin | (30-49) | g/L | 24 |
| Globulin | (17-29) | g/L | 12 |
| Bili Tot | (2-20) | umol/L | 204 |
| Bili Conj | (2-4) | umol/L | 82 |
| ALT | (5-80) | U/L | 79 |
| GGT | (10-150) | U/L | 38 |
| ALP | (75-435) | U/L | 890 |
| Ferritin | (10-120) | ug/L | 8750 |

Neonatal haemochromatosis

Girl child day of birth

| | | | |
|------------|----------|--------|-----------|
| Prot Total | (44-76) | g/L | 35 |
| Albumin | (30-49) | g/L | 21 |
| Globulin | (17-29) | g/L | 14 |
| Bili Total | (2-34) | umol/L | 91 |
| Bili Conj | (2-4) | umol/L | 69 |
| ALT | (5-80) | U/L | 57 |
| GGT | (10-150) | U/L | 77 |
| ALP | (75-435) | U/L | 149 |

TORCH infection

Causes of cholestasis in the younger than 2-month-old infant

Obstructive cholestasis

Biliary atresia
Choledochal cyst
Gallstones or biliary sludge
Alagille syndrome
Inspissated bile
Cystic fibrosis
Neonatal sclerosing cholangitis
Congenital hepatic fibrosis/
Caroli's disease

Genetic/metabolic disorders

1-antitrypsin deficiency
Tyrosinemia
Galactosemia
Bile acidopathies
Hypothyroidism/Panhypopituitarism
Neonatal Haemochromatosis.
Cystic fibrosis
Toxic/secondary
Parenteral nutrition-associated

Hepatocellular cholestasis

Idiopathic neonatal hepatitis

Infectious

Cytomegalovirus/Syphilis/TORCH(ES)

HIV

Bacterial infection/Sepsis

Urinary tract infection

7 y/o girl s/p choledochal cyst resection
(in way past). Now with belly pain.

| | | | |
|----------|----------|--------|-------------|
| Prot Tot | (63-83) | g/L | 73 |
| Albumin | (35-50) | g/L | 41 |
| Globulin | (20-36) | g/L | 32 |
| Bili Tot | (< 20) | umol/L | 6 |
| ALT | (5-45) | U/L | 12 |
| GGT | (5-40) | U/L | 11 |
| ALP | (95-390) | U/L | 154 |
| Lipase | (0-60) | U/L | 7600 |

3 year girl

| | | | |
|----------|----------|--------|------|
| Prot Tot | (50-78) | g/L | 76 |
| Albumin | (33-49) | g/L | 43 |
| Globulin | (20-36) | g/L | 33 |
| Bili Tot | (2-20) | umol/L | 50 |
| ALT | (5-45) | U/L | 107 |
| GGT | (5-40) | U/L | 394 |
| ALP | (95-390) | U/L | 445 |
| Lipase | (0-60) | U/L | 2286 |

Gall stone pancreatitis.

6 year boy belly pain a lot.

Date: 07/04/08 24/08/07

| | | | | |
|--------|--------|-----|------------|------------|
| Lipase | (0-60) | U/L | 965 | 396 |
|--------|--------|-----|------------|------------|

Chronic recurrent pancreatitis

18 month girl with
recurrent
screaming and
occasional
vomiting.



| | | | |
|--------|----------|--------|---------------|
| Lipase | (0-60) | U/L | 2830 |
| Trig | (< 1.70) | mmol/L | 115.00 |
| Chol | (< 4.50) | mmol/L | 5.72 |

Lipoprotein lipase deficiency

16 y/o girl with abd pain after bicycle wreck

| | | | | |
|---------|----------|-----|---|-------------|
| Amylase | (10-120) | U/L | H | 2652 |
| Lipase | (0-60) | U/L | H | 4570 |

06- 827424

589 children

Mean age - 9.2 years youngest 1 week

Male to female ratio -1.2.

Aetiologies (n = 589):

| | |
|----------------------|-------|
| idiopathic | (23%) |
| trauma | (22%) |
| structural anomalies | (15%) |
| multisystem disease | (14%) |
| drugs and toxins | (12%) |
| viral infections | (10%) |
| hereditary | (2%) |
| metabolic disorders | (2%). |

J Clin Gastroenterol. 2003 Aug;37(2):169-72.

Aetiology of pancreatitis

- Recurrence was reported in 9%, most with idiopathic and structural etiologies.
- Fatal outcome was described in 9.7% of the patients.

9 y/o girl:

Yellowish lesion R elbow. Present for years ?

MICROSCOPICAL EXAMINATION :

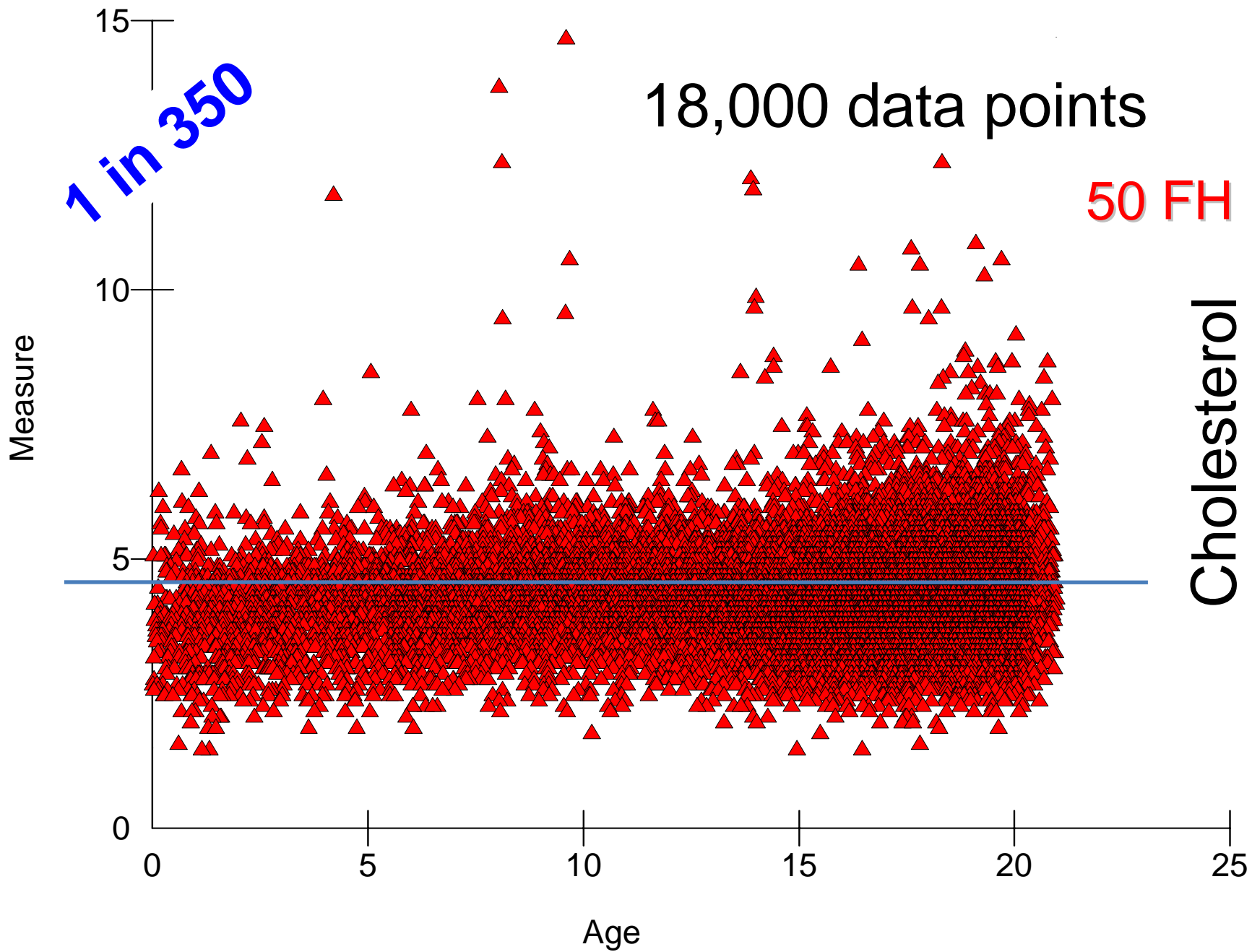
The dermis is infiltrated by numerous foamy histiocytes and at the deep aspect there is a distinct nodule composed of further foamy histiocytes intimately admixed with dense collagen fibres.

The appearances are those of a tuberous xanthoma. These lesions can be associated with hyperlipidaemia.

DIAGNOSIS :

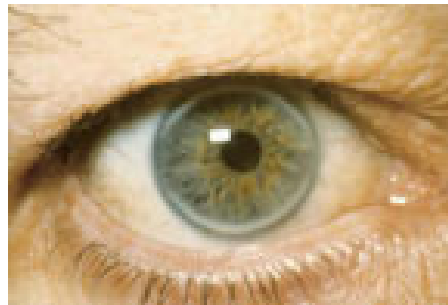
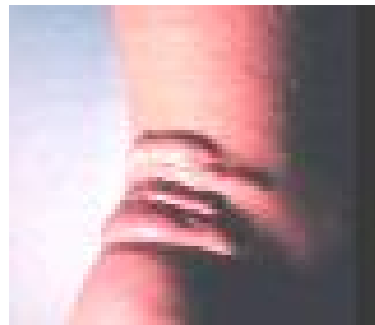
Punch biopsy, right elbow: Tuberous xanthoma.

| | | | | |
|-----------------|----------------------|---------------|----------|-------------|
| Chol | (< 4.50) | mmol/L | H | 9.24 |
| HDL Chol | (> 1.00) | mmol/L | | 1.37 |
| LDL Chol | (< 2.90) | mmol/L | H | 7.41 |
| Trig | (< 1.70) | mmol/L | | 1.01 |



Clinical features of Familial Hypercholesterolaemia

- Heterozygotes develop tendinous xanthomas, corneal arcus (50% by age 50 years), and xanthelasma.



- Otherwise minimal clinical clues
until chest pain



Morbidity & Mortality

- 50 % risk of myocardial infarction in men by age 50 years.
- 30% risk of myocardial infarction in women by age 60 years.
- Risk increases from age 20 years.

| AGE | Male Heterozygotes | | Female Heterozygotes | |
|----------|--------------------|----------------|----------------------|----------------|
| | Coronary Symptoms | Coronary Death | Coronary Symptoms | Coronary Death |
| 40 years | 20 | 5-10 | 3 | 0 |
| 50 years | 45 | 25 | 20 | 2 |
| 60 years | 75 | 50 | 45 | 15 |
| 70 years | - | 80 | 75 | 30 |

Primary Amenorrhoea 14 year girl

FSH IU/L H **110**

LH IU/L H **24**

FSH

LH

Follicular 3.0 - 10.0 4.0 - 30.0

Mid Cycle Peak 10.0 - 20.0 30.0 - 60.0

Post Menopausal > 20.0 > 30.0

Turner's Syndrome (XO)

1 in 2500 girls

Secondary Amenorrhoea 15 year girl

*Please note
report format*

| | | |
|--------|------|------|
| L.H. | IU/L | <0.1 |
| F.S.H. | IU/L | <0.1 |

| | L.H. | F.S.H. |
|-------------------|----------|----------|
| Follicular/Luteal | 1 - 12 | 4 - 10 |
| Postmenopausal | 10 - 60 | 25 - 110 |
| Mid-cycle Peak | 20 - 103 | 5 - 22 |

Anorexia Nervosa
3 in 1000 (90% girls)

Delayed Puberty 17 year boy

FSH (1.0-10.5) IU/L H **90**

LH (1.0-8.4) IU/L H **30**

Cancer survivor clinic (ALL)

16 year Gym Rat

L.H. **<0.1** mIU/ml

F.S.H. **0.2** mIU/ml

| | L.H. | F.S.H. |
|------|-----------|------------|
| Male | 2.0 - 8.0 | 2.0 - 10.0 |

Testosterone **45.8** nmol/L (8.5-30.0)

SHBG **6** nmol/L (15-70)

FAI **7633** (160-1190)

Exogenous hormone

Reproductive Issues

- Tanner Staging
- Developmental disorders

Hypergonadotrop(h)ic hypogonadism

| | Primary | Secondary |
|---------------|------------------|-------------------|
| Hypogonadism | HyperHypo | HypoHypo |
| Hypergonadism | HypoHyper | HyperHyper |

www.cysticfibrosis.org.au



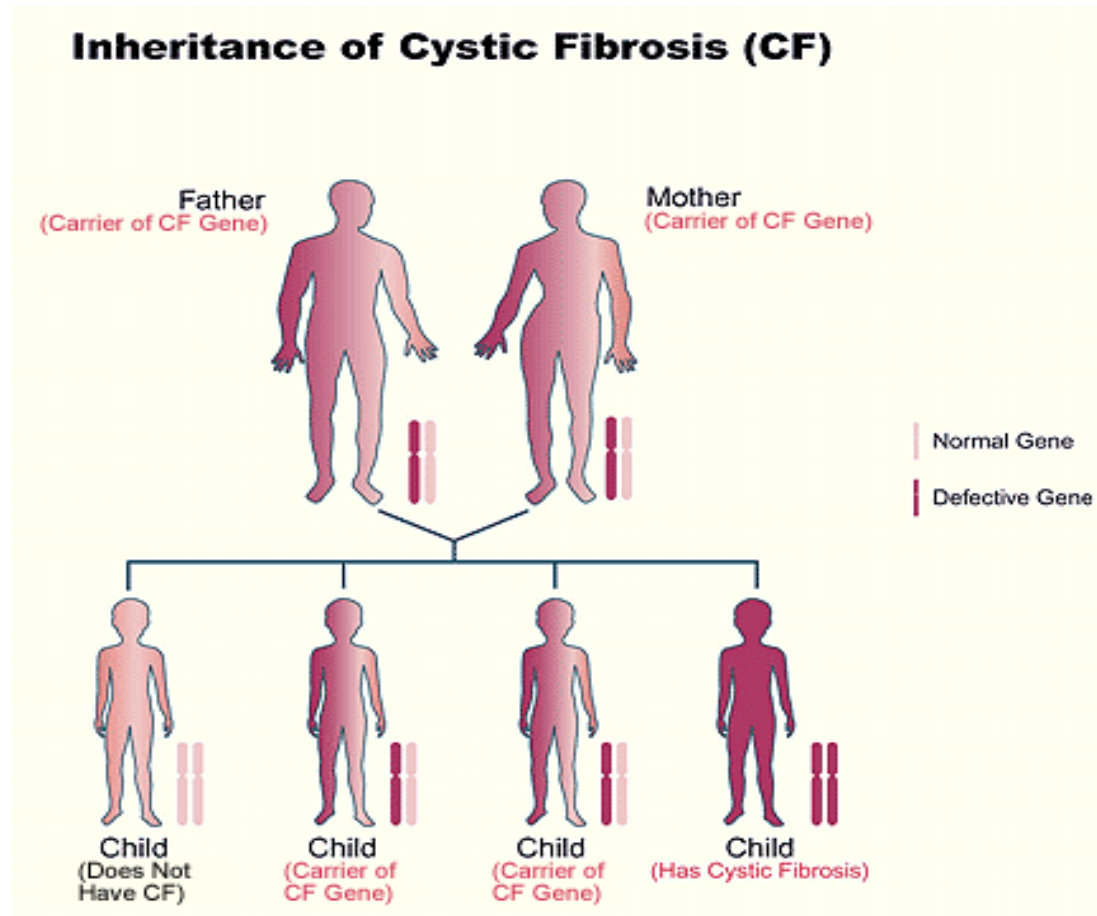
Sixty Five Roses

The CF support
group is 65 Roses.

<http://www.cff.org/>



CF is the most common autosomal recessive lethal disorder in children.



In SA, the frequency is about 1:2500 livebirths.

Newborn screening throughout Australia. Still about 5% missed by screening.

More than 70% of patients are diagnosed by age two.

CF in the lab

Pseudo-hypoaldosteronism.

Polyclonal gammopathy.

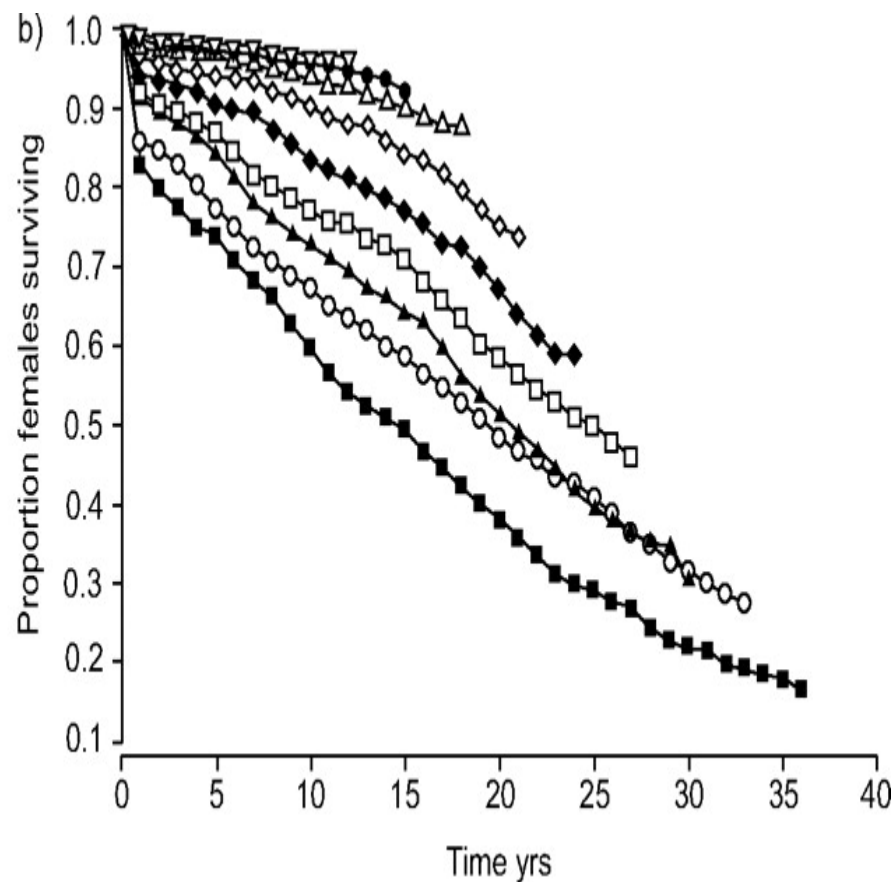
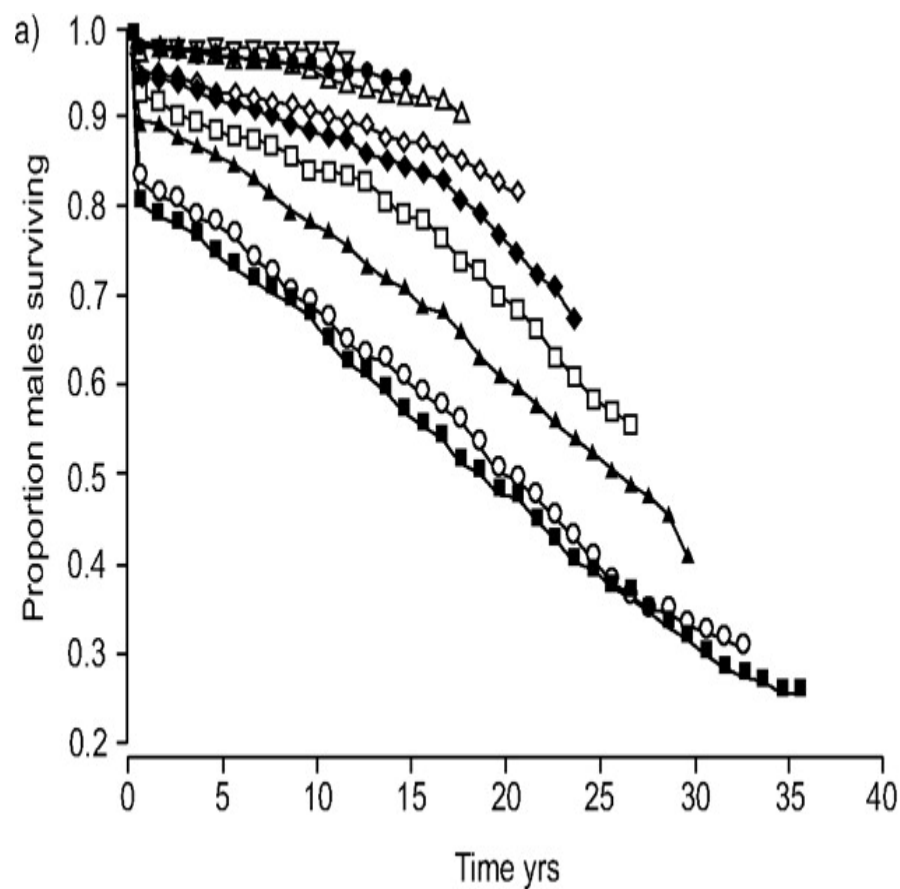
Respiratory acidosis & Metabolic alkalosis.

Fat soluble vitamin deficiency.

Pancreatitis.

Cholestasis.

Diabetes mellitus (ketosis resistant).



UK cystic fibrosis population. Proportion of a) males and b) females of each 3-yr cohort surviving until 2003. \square : 1968–1970; \diamond : 1971–1973; \triangle : 1974–1976; \circ : 1977–1979; \blacktriangle : 1980–1982; \square : 1983–1985; \blacklozenge : 1986–1988; \bullet : 1989–1991; 1992–1994.

Kid's Chemistry

- Infrequent disease
- Some special
- Some not
- All important
- Kids are people



Cases in Kids' Chemistry



MP Metz

AIMS ASM Adelaide 2009