

Dia 1


UMC St Radboud

Turner syndrome

Henri Timmers
internist-endocrinologist

Dept. of internal medicine
Section of endocrinology

UMC St Radboud
Turnerpolikliniek

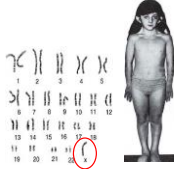


Dia 2

UMC St Radboud

Turner syndrome

1938
Henry Turner: description of clinical triad: -
- short stature
- 'sexual infantilism'
- dysmorphic abnormalities



1959
C.E. Ford: discovery of X chromosomal monosomy

1 APRIL 1959 ORIGINAL ARTICLES THE LANCET 711

A SEX-CHROMOSOME ANOMALY IN A CASE OF GONADAL DYSGENESIS (TURNER'S SYNDROME)
By C. E. FORD, K. W. JONES, and P. O. WARD

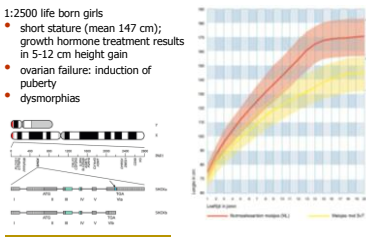
1958) or in cells from tissue cultures (Tjio and Paik 1958). In consequence the normal number of human chromosomes and their normal morphology are now reasonably well known. The subject of this report is a chromosomally negative case of Turner's syndrome whose karyotype is 45,X. In contrast to the normal karyotype only, instead of the normal number of 46, and whose sex-chromosomal constitution is determined to be XO...

Dia 3

UMC St Radboud

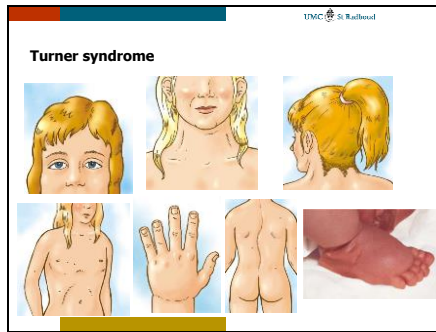
Turner syndrome

- 1:2500 live born girls
- short stature (mean 147 cm); growth hormone treatment results in 5-12 cm height gain
- ovarian failure: induction of puberty
- dysmorphias

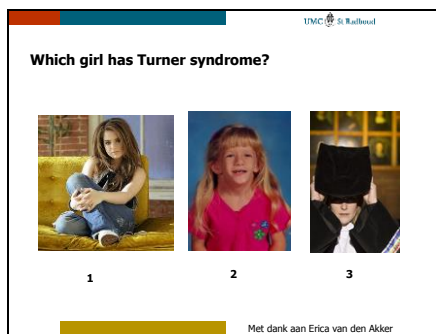


The graph shows height (cm) on the y-axis (100-180) and age (years) on the x-axis (0-20). It compares 'Turner syndrome (height %)' (red line) and 'Height for SD' (yellow line). The Turner syndrome curve is significantly lower than the normal SD curve, especially after age 10.

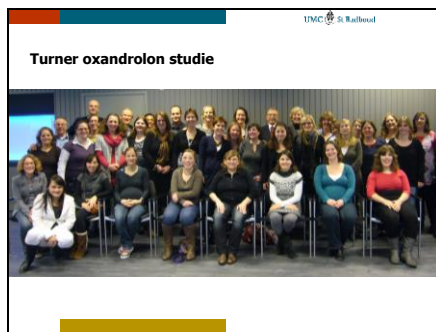
Dia 4



Dia 5



Dia 6



Dia 10


IMC St Radboud

Cardiovascular morbidity

Life expectancy - 13 years / cardiovascular mortality x 3

Structural abnormalities heart / aorta

- congenital: bicuspid aortic valve (15-30%), aortic coarctation (10-15%)
- acquired: aortic dilation / aneurysm (25%) and dissection



1. normal aortic valve
2. bicuspid aortic valve
3. aortic coarctation

4. aortic dilation / aneurysm


Dia 11

IMC St Radboud

Cardiovascular morbidity

Increased risk of atherosclerosis

- hypertension
 - ✓ 50% of young adults
 - ✓ chance of dissection in case of aortic dilation / bicuspid aortic valve (treatment goal SBP <120 mmHg)
- diabetes mellitus type 1 x1.1, type 2 x3.5
- dyslipidemia 40-50% of young adults

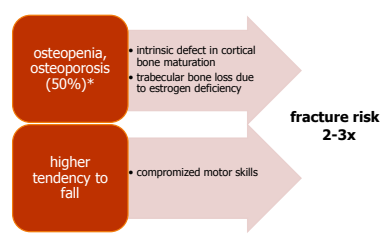


4. aortic dilation / aneurysm

Dia 12

IMC St Radboud

Osteoporosis and fractures



osteopenia, osteoporosis (50%)*

- intrinsic defect in cortical bone maturation
- trabecular bone loss due to estrogen deficiency

higher tendency to fall

- compromized motor skills


fracture risk 2-3x

*DEXA underestimates BMD in short stature

TMG St. Radboud

Conclusions

- Besides short stature and ovarian failure, patients with Turner syndrome are prone to a wide range of morbidity throughout life with a large phenotypic variation
- This requires multidisciplinary care with standardized screening and careful transition from pediatric into adult medical care



The composite image consists of two parts. On the left, a hand is shown holding a globe, symbolizing global care or a wide range of phenotypic variation. On the right, a circular diagram features a central image of a person with the letters 'U' and 'N' overlaid. Surrounding this central image are six boxes representing medical specialties: 'prenatal endocrinologist' at the top, 'cardiologist' on the right, 'psychologist' at the bottom right, 'other' at the bottom, 'obstetrician/gynecologist' on the left, and 'gynecologist' at the top left.
