Seckel syndrome: an overdiagnosed syndrome

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SUMMARY Five children in whom a diagnosis of Seckel syndrome had previously been made were re-examined in the genetic unit. One child had classical Seckel syndrome, a sib pair had the features of the syndrome with less severe short stature, and in two children the diagnosis was not confirmed. Seckel syndrome is only one of a group of low birth weight microcephalic dwarfism and careful attention should be paid to fulfilment of the major criteria defined by Seckel before the diagnosis is made. There remains a heterogeneous group of low birth weight microcephalic dwarfism yet to be defined.

Seckel¹ first defined the syndrome as severe intrauterine growth retardation (IUGR); severe short stature which is proportionate; severe microcephaly; a 'bird headed' profile with a receding chin and forehead and large beaked nose; mental retardation; and various associated congenital anomalies, which is inherited in an autosomal recessive manner. Although an excessively rare condition, the name is known to most paediatricians and it appears to be a condition that is overdiagnosed.

Majewski and Goecke² reviewed the 60 published cases of the syndrome and found only 17 who fulfilled the original diagnostic criteria. They adopted strict criteria and found that the average birth weight at term was 1543 g (range 1000 to 2055 g). The approximate mean postnatal growth retardation in 16 of the 17 was -7.1 SD (range -5.1 to -13.3). The mean occipitofrontal head circumference (OFC) was -8.7 SD (range -4.1 to -14.3 SD); in half the OFC was as retarded as height and in half more retarded than height. All were mentally retarded, nearly half with an IQ of less than 50. In addition, they compiled a list of the variable associated abnormalities of the syndrome.

The purpose of this report is to document further evidence for the heterogeneity of low birth weight microcephalic dwarfism and to present a sib pair with Seckel syndrome who are taller than previously reported cases. Useful diagnostic features among the associated abnormalities are also illustrated. Detailed anthropometric measurements have been recorded (appendix 1 and 2).

Case reports

CASE 1

This patient, a male aged 14 years, was the first born

FIG 1 Extract of growth records of cases 1 to 5. (Standards of Tanner et al. Arch Dis Child 1966;41:613.)

FIG 2 Side view (a), front view (b), hands (c), and standing posture (d) of case 1.

of twins; the co-twin died at three days of age of a heart defect. The birth weight of case 1 was 1701 g. Further information about the pregnancy and perinatal period was not available; the child was in institutional care for the first 11 years of life and was then fostered. He had a cleft palate which was repaired and suffered from frequent upper respiratory tract infections while in institutional care. Orthodontic treatment and tooth extractions had been required for crowded teeth. The mental age was assessed as 5 years 9 months at a chronological age of 9 years and he attends a special school for the educationally subnormal. Only one previous height measurement (supplied by his foster mother) was available (fig 1). On examination he was very amiable and cooperative. The height was 127.4 cm (-5.1 SD), weight 20.5 kg, and head circumference

44.5 cm (-6.6 SD). The short stature was proportionate, that is, the leg to trunk length ratio was normal (appendix 1). There was micrognathia, a receding forehead, a large curved nose, and small, lobeless ears (fig 2a, b).

The face was asymmetrical, the teeth crowded, and the palpebral fissures small. The eyes appeared widely spaced but there was telecanthus and the interpupillary distance was proportionate to face width (appendix 2). The frontal hairline was V shaped. The total hand length was small but compatible with height and the fingers were short relative to palm length (fig 2c, appendix 1). There was camptodactyly involving the proximal interphalangeal joints. The feet were small for age but relatively large for height (appendix 1). There was pes planus, the toes were short and tapered with a

gap between the right first and second toes, and there was a left hallux valgus. The toes could not be flexed. The heads of the radii were clinically and radiologically dislocated. There was mild flexion at the hips and knees, giving a characteristic standing posture (fig 2d). The cardiovascular system and genitalia were normal and there was no hirsutism.

CASE 2

This patient, a male aged 15 years 8 months, was the first child of healthy unrelated English parents. The fetus was small for dates throughout pregnancy. Delivery at 37 weeks was normal and there was no birth asphyxia. Birth weight was 1980 g. A congenital right talipes equinovarus was treated surgically. Early feeding and respiratory problems led to care in the hospital special nursery for the first six weeks of life. From the ages of 5½ months to 5 years there were frequent attacks of bronchitis. Insulin dependent diabetes mellitus developed at 12 years. He wears hearing aids and has had dental extractions and orthodontic treatment for crowded teeth. He

attends a special school for the severely educationally subnormal and has behaviour problems. Previous height measurements supplied by the local hospital are charted in fig 1. The maternal and paternal heights were 152 cm and 189 cm respectively. Mid-parental height correcting for maternal sex was 176.75 cm, which is just above the 50th centile for an adult male.

On examination, the height was 150.2 cm (-3.3 SD), weight 43.1 kg, and head circumference 44.2 cm (-7.3 SD). The short stature was proportionate (appendix 1). The face was round and asymmetrical, the nose large and curved, the chin small, the forehead receding, and the ears small and lobeless (fig 3a). The palate was high and narrow, the teeth crowded, and the palpebral fissures small with telecanthus. The interpupillary distance was proportionate to face width (fig 3b, appendix 2). The hands and feet were proportionate to height but with relatively short fingers (appendix 1). There was clinobrachydactyly of the fifth fingers (fig 3c) and clinodactyly of the second left toe and a wide gap

FIG 3 Side view (a), front view (b), hands (c), and x-ray of hands (d) of case 2.

between the first and second toes. Previous hand x-rays showed a pseudoepiphysis at the base of the second and fifth metacarpals and fusion of the trapezium and scaphoid bones and of the triquetrum and lunate bones (fig 3d). A previous chest x-ray revealed 12 ribs. The heads of the radii were clinically and radiologically dislocated. Slight fixed flexion of the hips and knees produced the same standing posture as in case 1.

CASE 3

This patient, a female, the only sib of case 2, was seen at 12 years 5 months. She was born at term, weighing 1814 g, and remained in hospital for a month to gain weight. She had 'bronchitis' at 7 weeks. She has had orthodontic treatment for crowded teeth. A Stanford-Binet assessment at a chronological age of 3 years 11 months showed a mental age of 2 years 9 months. She attends a school for the educationally subnormal and is of a particularly outgoing and happy disposition. Previous height and weight measurements supplied by the local hospital are charted in appendix 1. On examination the height was 132.4 cm (-3.4 SD), weight 30.2 kg, and head circumference 40.9 cm (-9.4 SD). The short stature was proportionate

(appendix 1). Fig 4a and b show the large curved nose, receding forehead and chin, small lobeless ears, asymmetrical face, and crowded teeth. The palpebral fissures were small with telecanthus and the interpupillary distance was normal relative to face width (appendix 2). The hands and feet were proportionate to height and the fingers proportionate to palm length (appendix 1), apart from brachydactyly of the fifth fingers (fig 4c). There was clinodactyly of the second left toe and a gap between the first and second toes. The heads of the radii were clinically dislocated, with inability to fully supinate the forearms and a tendency to hold the elbows flexed. The same stance as in cases 1 and 2 was present. There was no clitoromegaly or hirsutism. Fig 4d shows the sibs at a younger age when their facial features were less angular and the eyes appeared to bulge, particularly in the girl.

CASE 4

This patient, a male aged 7 years 8 months, was born of the first pregnancy of healthy unrelated English parents. He was born at term of a normal delivery. Birth weight was 1500 g, length 41 cm, and head circumference 29 cm. Seckel syndrome was first diagnosed at 5 months of age when he weighed 2.5 kg

FIG 4 Side view (a), front view (b), and hands (c) of case 3. (d) Sibs, cases 2 and 3, at 5 and 2 years respectively.

TABLE Summary of manifestations.

	Seckel syndrome			Published cases ²	Undiagnosed dwarfism	
	Case 1	Case 2	Case 3		Case 4	Case 5
Intrauterine growth retardation (bw <2055 g at term)	+	+	+	17/17	+	+
Short stature						
Height -5 SD	+			16/17	+	
Height -3 SD		+	+			+
Proportionate trunk to leg length	+	+	+	17/17	-	+
Microcephaly	+	+	+	17/17	+	+
Mental retardation	+	+	+	17/17	+	+
Face						
Micrognathia	+	+	+	17/17	-	-
Large curved nose	+	+	+	17/17	-	-
Receding forehead	+	+	+	17/17	-	-
Asymmetry	+	+	+	NR	-	-
Eyes						
Small palpebral fissures	+	+	+	NR	+	-
Telecanthus	+	+	+	NR	-	+
Bulging eyes	-	+	+	10/16 large eyes	-	-
Antimongoloid slant	-	(at a young age)	-	7/11	+	+
Palate						
High arched	-	+	-	4/10	+	+
Cleft	+	-	-	2/17	-	-
Ears						
Small	+	+	+	NR	+	-
Lobeless	+	+	+	10/12 'dysplastic'	-	-
Teeth						
Crowded	+	+	+	NR	-	-
Malocclusion	+	+	+	NR	+	+
Skeleton						
Hand abnormalities*	+	+	+	8/8 clinodactyly V	-	+
Foot abnormalities*	+	+	+		-	-
Dislocated heads of radii	+	+	+	3/6	-	+
Hips: fixed flexion	+	+	+	5/9 hip 'dysplasia'	-	-
Hips: congenital dislocation	-	-	-		-	+
Knees: fixed flexion	+	+	+	NR	-	-
Dislocation	-	+	-	NR	-	-
Genitalia						
Cryptorchidism	-	-		3/4	+	
Clitoromegaly			-	3/7		-

*See text for specific abnormalities. NR=not recorded.

and was 50.5 cm in length, with a head circumference of 34 cm. He had bilateral radical mastoidectomies at 5 years. Although early motor milestones were normal, intellectual handicap became apparent and he attends a school for the severely educationally subnormal. Previous measurements from the hospital file are given in fig 1. It was the mother who noticed dissimilarities between him and other children with Seckel syndrome. The maternal and paternal heights were 156 cm and 181.5 cm respectively; mid-parental height correcting for maternal sex was 175.0 cm which is on the 50th centile for an adult male. There was one sib, a normal girl. On examination, the height was 95.7 cm (-5.1 SD), weight 11.0 kg, and head circumference 42.5 cm (-7.3 SD). The short stature was not proportionate as the trunk was relatively short compared to the

legs (appendix 1). Fig 5a shows the child at 5½ years with his 3½ year old sister. The nose was not unduly prominent or curved and the forehead and chin did not recede (fig 5b, c). The palpebral fissures were small and appeared closely spaced but the interpupillary distance was proportionate to the very narrow face width (appendix 2). The ears were normally shaped and the palate was high arched. The hands and feet were proportionately small and the fingers were in proportion to palm length and normally shaped (appendix 1). The elbows, hips, and knees were normal. There was bilateral cryptorchidism and no hirsutism. A dental report indicated that several teeth were absent and a class III malocclusion was present. A chromosome karyotype (G banded) was normal (46,XY) and there were no increased numbers of sister chromatid

FIG 5 (a) Case 4 at 5½ years with normal sister aged 3½ years and parents. (b) Side view and (c) front view of case 4.

exchanges (SCE) with bromodeoxyuridine in the culture.

CASE 5

This patient, a female aged 7 years 9 months, was born of the first pregnancy of healthy unrelated English parents. There was one sib, a girl, who had had a right congenital dislocated hip but was otherwise healthy. Delivery was at term after a normal pregnancy. The birth weight was 1899 g and length 40.6 cm. Bilateral hip dislocations were diagnosed at six months and were corrected with splinting. Repeated chest infections occurred during infancy, and eczema and asthma were diagnosed at three years. She attends a school for the educationally subnormal and is hyperactive. Previous height measurements were not available. On examination she was overactive with mild truncal and limb ataxia. The height was 107.3 cm (−2.9 SD), weight 20.6 kg (10th

centile), and head circumference 46.7 cm (−4.0 SD). The trunk to leg length proportions were normal (appendix 1). The nose was large but not curved, the chin and forehead were normal, and the ears large and simple with small lobes (fig 6). There was a mild antimongoloid eye slant and a left convergent strabismus and telecanthus (appendix 2). The palate was high arched. The hands and feet were proportionally small and the fingers proportionate to palm length, apart from clinodactyly of the fifth fingers (appendix 1). The heads of the radii were clinically dislocated and the elbows could not be fully supinated. The Achilles tendons were shortened so that the heel could not be placed flat on the floor. Chromosome karyotype (G banded) was normal (46,XX) and there were no increased numbers of SCE on chromosome culture with bromodeoxyuridine. Immunoglobulins and alphafetoprotein were normal.

Discussion

Diagnostic problems arise when children manifest many, but not all, the classical features of Seckel syndrome. Our approach has been to evaluate all five cases on the same day, comparing each child with case 1, who presents a classical case of Seckel syndrome, meeting the most stringent criteria. In addition to intrauterine growth retardation, proportionate short stature, microcephaly, and mental retardation, the sib pair (cases 2 and 3) had a strikingly similar facial appearance to case 1, together with small lobeless ears (cases 1, 2, and 3); small hands (with shortening of the fingers relative to the

FIG 6 Case 5.

palm length in cases 1 and 2); small feet with a wide gap between the first and second toes (cases 1, 2, and 3); dislocation of the heads of the radii (cases 1, 2, and 3); and a characteristic stance with fixed flexion at the hips and knees. On the basis of these discrete dysmorphic features shared in common with case 1 we concluded that the sib pair do have Seckel syndrome and that therefore a height above -5 SD does not exclude the diagnosis, as suggested by Majewski and Goecke.² Two sibs originally reported by Frijns and Van den Berghe,³ excluded by Majewski and Goecke as being too tall for the Seckel syndrome, are similar to our sibs. They are a female aged 23 years and a male aged 14 years with birth weights at term of 1400 g and 1700 g respectively, heights of -4.7 SD and -2.1 SD respectively, and microcephaly and severe mental retardation. The boy's face is less typical and he is mildly retarded.³

The table compares the salient features of our cases 1, 2, and 3 with the 17 cases drawn from published reports by Majewski and Goecke.² It is seen that we confirm several previously recognised associations as well as draw attention to several others not previously emphasised. Facial asymmetry, not emphasised by Majewski and Goecke,² is present in our three cases and in the sibs described by Harper *et al.*⁴ 'Large' eyes have been commonly reported (table) and variously described as 'protruding',⁴ 'proptosed',⁵ and 'bulging',⁶ suggesting that the globes are large relative to the bony orbits. Ocular measurements have not previously been reported. Our cases 1, 2, and 3 have short palpebral fissures with marked telecanthus; the appearance of bulging eyes was seen in the sibs only at a young age. Widely spaced eyes have been previously reported. In our cases, interpupillary distance is proportionate to the narrow face. Dysplastic ears are common; the abnormality in about half the published cases is lobeless ears, as in our three cases.^{1 4 7-9} The dental overcrowding and malocclusion presumably relate to the small jaws. Enamel hypoplasia, another commonly reported feature, is absent in our cases since it mainly affects the first dentition.⁵ Abnormalities of the hands and feet, and joints, such as the elbows, hips, and knees, are an important manifestation of the syndrome. The most common abnormality of the hands is clinodactyly of the fifth fingers (table). Abnormal finger flexion creases, as in our case 1, have been reported in four cases.^{1 4 9} General shortening of fingers relative to palm length (cases 1 and 2) has not previously been reported in Seckel syndrome. Carpal bone fusion (case 2) occurred in Seckel's case 1; the pseudoepiphyses (particularly of the second finger) are a non-specific finding. The wide gap between the first and second

toes of our cases 1, 2, and 3 was reported by Seckel¹ and Harper *et al.*⁴ Dislocation of the heads of the radii is characteristic and combined with fixed flexion at the hips and knees produces a typical standing posture, noted previously by de la Cruz.⁷

Our case 4 does not have Seckel syndrome. Similarities include very low birth weight, severe short stature, microcephaly, and mental retardation. Not consistent are the relatively small trunk to leg length ratio, the normally proportioned forehead and mandible, and the small but otherwise normal extremities and joints (appendix 1). The eye measurements are proportionate to the narrow face width in contrast to the Seckel group in whom there is marked relative widening of the inner canthal distance. The high palate and malocclusion may relate to an extremely narrow face, and cryptorchidism is non-specific. The features are not those of other low birth weight microcephalic dwarfisms, for example Cornelia de Lange, Russell-Silver, Dubowitz, or Fanconi syndromes. Bloom syndrome is excluded by normal SCE.¹⁰ The osteodysplastic primordial dwarfism types I, II, and III described by Majewski *et al.*¹¹⁻¹³ are excluded, as the craniofacial abnormalities in these conditions are the same as in Seckel syndrome and, in addition, in type II the limbs are relatively short. The cause of the problems of case 4 remains unknown.

Case 5 has a disorder different from both Seckel syndrome and from case 4, with low birth weight, microcephaly, and mental retardation, but in contrast to Seckel syndrome moderate short stature, a normally proportioned chin and forehead, and relatively large ears. The high palate, downward slanting palpebral fissures, telecanthus, strabismus, clinodactyly, and dislocated radii and hips are reminiscent of Seckel syndrome. Ataxia is not a feature of Seckel syndrome. Absence of telangiectasia and normal immunoglobulins and alpha-fetoprotein exclude ataxia-telangiectasia. The precise diagnosis for case 5 is unknown.

In conclusion, Seckel syndrome is only one of a group of low birth weight microcephalic dwarfisms and can be overdiagnosed. There is a continuing need to report cases that have been documented carefully, so that the full range of expression of this autosomal recessive syndrome can be delineated.

Addendum

After this report was submitted, another child with Seckel syndrome was seen, who is briefly described since he is much younger than our other cases. He was an 18 month old boy who had a birth weight at term of 2.18 kg, a height of -5.2 SD with proportionate body ratios, a head circumference of -4.6

FIG 7 See addendum.

SD, and developmental retardation. The nose was large with a prominent nasal bridge and the chin small, but the forehead was less receding compared to the older children with Seckel syndrome (fig 7). He demonstrated several features emphasised in this report, namely facial asymmetry, narrow palpebral fissures, telecanthus, and small ears (which did have lobes). In addition there was clinodactyly of the fifth fingers and third toes bilaterally, tapered fingers, and a dislocated right radial head clinically and on x-ray. There were pseudoepiphyses at the bases of the second metacarpals and no ossification of the middle phalanges of the toes. The testes were undescended. Further details are available from the authors on request.

Although there is little doubt that this child does have Seckel syndrome, he demonstrates that it is probably more difficult to make the diagnosis in the younger child before the typical facial profile has fully evolved.

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APPENDIX 1 Anthropometric measurements.

	Case 1	Case 2	Case 3	Case 4	Case 5
Age (y)	14	15 8/12	12 5/12	7 8/12	7 9/12
Birth weight at term (g)	1701	1984	1914	1500	1899
Height (cm)*	127.4 (-5.1SD)	150.2 (-3.3SD)	132.4 (-3.4SD)	95.7 (-5.1SD)	107.3 (-2.9SD)
Weight (kg)*	20.5 (<3rd centile)	43.1 (<3rd centile)	30.2 (<3rd centile)	11.0 (<3rd centile)	20.6 (10th centile)
Head circumference† (cm)	44.5 (-6.6SD)	44.2 (-7.3SD)	40.9 (-9.4SD)	42.5 (-7.3SD)	46.7 (-4.0SD)
Sitting height* (cm)	70.4 (-4.0SD)	79.8 (-3.3SD)	69.5 (-3.7SD)	48.8 (-7.1SD)	60.3 (-1.3SD)
Subischial leg length, ie height less sitting height* (cm)	57.0 (-4.8SD)	70.4 (-2.5SD)	62.9 (-2.2SD)	46.9 (-2.5SD)	47 (-2.3SD)
SD subischial leg length less SD sitting height*, ie leg to trunk ratio	-0.8 (Proportionate)	+1.0 (Proportionate)	+1.5 (Proportionate)	+4.6 (Legs relatively long)	-1.0 (Proportionate)
Triceps skinfold‡ (mm)	3.0 (<3rd centile)	17.0 (90-97th centile)	8.2 (10-25th centile)	8.1 (50th centile)	5.8 (3-10th centile)
Subscapular skinfold‡ (mm)	4.0 (<3rd centile)	10.2 (75th centile)	6.5 (10-25th centile)	4.6 (25-50th centile)	4.4 (10-25th centile)
Biacromial diameter†† (cm)	26.1 (-4.1SD)	32.6 (-2.1SD)	29.1 (-2.3SD)	21.8 (-4.5SD)	25.6 (10th centile)
Bi-iliac diameter†† (cm)	19.9 (-2.8SD)	25.4 (25th centile)	23.1 (10-25th centile)	14.4 (-4.7SD)	17.9 (3-10th centile)
Upper arm circumference‡‡ (cm)	16.0 (-3.3SD)	26.1 (-0.4SD)	18.9 (-1.2SD)	15.3 (-1.6SD)	17.7 (-0.6SD)
Palm length§ (cm)	8.6	8.9	8.4	6.1	7.1
Middle finger length§ (cm)	4.6 (-4.3SD)	5.8 (-4.8SD)	6.2 (-1.8SD)	4.8 (-2.8SD)	5.3 (-1.5SD)
Total hand length§ (cm)	13.2 (-3.8SD)	14.7 (-4.2SD)	14.6 (-1.7SD)	10.9 (-3.8SD)	12.4 (-1.9SD)
Middle finger % total hands	35%	40%	43%	44%	43%
Foot length§§ (cm)	20.5 (-3.1SD)	21.3 (-4.3SD)	19.3 (-3.1SD)	14.9 (-5.1SD)	16.8 (-2.5SD)

*Standards of Tanner JM, Whitehouse RH, Takaishi M. *Arch Dis Child* 1966;41:452-613 and 1976;51:170-9.

†Charts of Tanner JM, Whitehouse RH. Standard deviation scores are calculated only if the values are distributed evenly about the mean. Otherwise, centiles are given. The 3rd and 10-25th centiles correspond to -1.881, -1.282, and -0.675 SD respectively.

‡Standards of Snyder RG *et al*. Anthropometry of infants, children and youths to age 18 for product safety design. Consumer product safety division, Michigan, 1977.††Standards of Fingold M, Bossert WH. Normal values for selected physical parameters: an aid to syndrome delineation. *Birth Defects* 1974;X:13.

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APPENDIX 2 Facial measurements.

	Case 1	Case 2	Case 3	Case 4	Case 5
Face width* (cm)	11.8 (−1.4SD) [(11.8) (−1.4SD)]	11.4 (−3.2SD) 7.1 (−3.1SD) for 14 y)	10.5 (−2.5SD) 6.6 (−3.5SD)	9.3 (−4.6SD) 5.1 (−5.5SD)	11.1 (0.6SD) 6.4 (−3.1SD)
Outer canthal distance† (cm)	3.6 (97th centile) [(7.2) (−2.9SD)]	2.9 (25–50th centile) for 14 y)	2.6 (3–25th centile) 20 (Normal range 23–33)	2.1 (3–10SD)	2 (3–25th centile) 19 (Normal range 19–29)
Inner canthal distance† (cm)	17 (Normal range 23–33)	21 (Normal range 23–33)	4.6 (−2.8SD) for 14 y)	3.6 (−5.0SD)	4.3 (−2.8SD)
Palpebral fissure length‡ (mm)	5.0 (−1.9SD)	4.8 (−2.1SD) for 14 y)	4.2 (−3.2SD)	3.9 (−4.0SD)	5.7 (50th centile) 1.45
Interpupillary distance† (cm)	4.3 (−3.0SD)	1.3 (1.15)	1.2	1.0	10.0 (−1.4SD) Mouth open
Ear length† (cm)	1.3 (1.15)	9 (−4.8SD)	9.2 (−1.4SD)	8.4 (−1.8SD)	2.8
Philtrum length (cm)	11.5 (+0.8SD) Mouth open	3.4	3.0	2.6	4.2 (+1SD) Mouth open
Lower face height* (cm)	2.5 (2.45)	4.3 (−0.8SD)	3.2 (−2.2SD)	2.6 (−3SD)	
Nose width (cm)	3.5 (−2SD) (3–4) (−2.2SD) Mouth open)				
Mouth width (cm)					

Facial measurements were taken using a new photographic technique being developed by this department (MA Patton, 1984, personal communication). As standards for this technique are not yet available, values were compared with various standards derived from measurements made with conventional sliding calipers, as indicated. As a result there is likely to be a small error in the standard deviation scores; the values in brackets for case 1 were taken with calipers and provide comparison. It is planned to review the data when photographic standards are available.

*Standards of Snyder RG, *et al.* †Standards of Feingold M, Bossert WH (see appendix 1). ‡Normal ranges of palpebral fissure length given by Fox SA. The palpebral fissure. *Am J Ophthalmol* 1966;62:73.