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## An Anthropometric Study of 38 Individuals With Prader-Labhart-Willi Syndrome

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### Abstract

Weight, height, sitting height, and 24 other anthropometric variables (5 body circumferences, skinfolds at 7 sites, 4 head dimensions, and 8 hand and foot measurements) were obtained on 38 Prader-Labhart-Willi syndrome (PLWS) individuals (21 with apparent chromosome 15 deletions and 17 nondeletion cases) with an age range of 2 weeks to 38½ years. More than half of these individuals were measured on more than one occasion. The measurements confirmed the presence of short stature, small hands and feet, obesity, and narrow bi-frontal diameter in PLWS. No differences were found for the anthropometric measurements between the 2 chromosome subgroups. Inverse correlations were produced with linear measurements (eg, height, hand and foot lengths) and age, which indicated a deceleration of linear growth relative to normal individuals with increasing age.

### Keywords

chromosome 15 deletion; correlation studies

### INTRODUCTION

The Prader-Labhart-Willi syndrome (PLWS), generally sporadic in occurrence, is characterized by infantile hypotonia, early childhood obesity, mental deficiency, short stature, small hands and feet, and hypogonadism. The incidence of PLWS has been estimated at one in 25,000 live-births and accounts for about 1 % of all mentally retarded persons [Zellweger and Soper, 1979]. The cause of this syndrome is not clear, although a chromosome deletion has been found recently in about one-half of PLWS individuals [Ledbetter et al, 1982; Butler and Palmer, 1983; Mattei et al, 1984].

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Few, if any, comprehensive anthropometric studies of PLWS patients have been conducted. Longitudinal studies of PLWS individuals are also rare. Herein, we report results from anthropometric studies of 38 PLWS individuals with emphasis on chromosome findings.

## MATERIALS AND METHODS

Subjects included in this study were diagnosed as having PLWS on the basis of infantile hypotonia, hypogonadism, delayed psychomotor development and/or mental retardation, early childhood obesity, small hands and feet, and short stature. The sample included 22 males and 16 females ranging in age from 2 weeks to 38½ years with a mean age of 13 1/12 years.

All of the anthropometric measurements were made by one of the authors (F.J.M.) according to standard techniques as presented by Weiner and Lourie [1969]. The longitudinal data were collected over a period of 3½ years.

The measurements consisted of the following: weight, height (or length), sitting height, total hand length, palm length, middle finger length, hand breadth, wrist breadth, ankle breadth, total foot length, foot breadth, head length, head breadth, minimum frontal diameter, maximum frontal diameter, chest circumference, waist circumference, upper arm circumference, calf circumference, head circumference, triceps skinfold, subscapular skinfold, forearm skinfold, supra-iliac skinfold, abdomen skinfold, thigh skinfold, and medial calf skinfold. Up to the age of 2 years, length was measured using a horizontal calibrated board in supine position, and thereafter height measurements were taken. Skinfold measurements were obtained to the nearest half millimeter with a Lange Skinfold Caliper. Circumferences were obtained to the nearest millimeter with a steel tape. Weight and height were measured using a balanced-beam scale and anthropometer, respectively.

Z scores were computed using normative measurements to control for age and sex effects. Z scores were calculated from the following formula:  $Z = \frac{X - \bar{X}}{SD}$  where SD is standard deviation of the normal control;  $\bar{X}$  is the mean of the normal control; and X is the measurement of the subject. Normative values were obtained from several sources [National Center for Health Statistics, 1970, 1972, 1974a,b, 1977; Tanner and Whitehouse, 1975; Snyder et al, 1977; Farkas, 1981].

High-resolution chromosome analysis was performed on the 38 PLWS individuals. Twenty-one PLWS individuals were identified with an apparent deletion of the proximal long arm of chromosome 15 and normal chromosome findings in the remaining 17 individuals.

## RESULTS

Table I shows clinical data of the 38 patients. Z score data are shown in Table II for 15 anthropometric measurements. It was impossible to calculate Z scores for all measurements because adequate normative data do not exist. A summary of PLWS anthropometric measurements is shown in Table III.

Longitudinal anthropometric measurements were obtained on several PLWS individuals. Figures 1–9 show the growth curves for weight, height, sitting height, hand length, finger length, hand breadth, foot length, and foot breadth. Most males and females were overweight, but weight loss was achieved with a controlled diet in some individuals (Figs. 1, 2). More females than males were below the 5th centile for height, particularly from 2 to 16 years. Most females and males were in the normal range for sitting height, indicating that the short stature (common in this syndrome) may be related to a shorter lower body segment compared with upper body in both sexes. Most females had hand and foot length at or below the 5th centile while males were grouped between the 50th centile and at or below the 5th centile. Similar findings were observed with middle finger length, foot breadth, and hand breadth. No differences were found in the anthropometric measurements between the 2 chromosome subgroups.

Although the anthropometric measurements were standardized for age and sex, several of these measurements were inversely correlated with age [Ward and Meaney, 1984]. There was no difference between deletion and nondelation individuals with age effect.

## DISCUSSION

No difference in anthropometric measurements was found between the 2 PLWS subgroups. A larger survey of PLWS patients and first-degree relatives and longitudinal studies of PLWS individuals are needed before a final conclusion may be reached with regard to anthropometric differences between the 2 chromosome subgroups or between sexes.

Although anthropometric measurements were standardized for age and sex, several measurements were inversely correlated with age [Ward and Meaney, 1984]. For example, correlations between anthropometric Z score variables and age were negative for the 19 hand bones, height, sitting height, hand and foot measurements, and head circumference and length. No relationship was found between age and Z scores for weight, head breadth, minimum frontal breadth, skinfolds, and body circumferences. It has been suggested elsewhere [Ward and Meaney, 1984] that these relationships may represent a relative deceleration in linear growth with increasing age in PLWS individuals as compared with normal individuals. The origins of the short stature and other decreased linear measurements in PLWS patients are not clear. Attempts to treat PLWS individuals with anabolic steroids have met with limited success in improving their overall growth and stature [Nugent and Holm, 1981]. Research to understand growth deficiency in PLWS is underway [Nugent and Holm, 1981].

Measurements of fatness such as skinfold thicknesses, particularly over the triceps, may reflect a predisposition toward development of excessive fatness or obesity in PLWS individuals before this is evident by physical examination [Ward and Meaney, 1984]. Early postnatal growth measurements of height and weight in combination with an assessment of fatness development may provide an improved means of PLWS diagnosis in infants in which the clinician has observed characteristics such as hypotonia, hypogonadism, low birth weight, and feeding problems.

Earlier diagnosis of PLWS might eventually improve the chances for more effective dietary management. While it may be of some benefit to use growth in height and weight in attempting to assess the development of obesity in suspected PLWS patients, the results from this study suggest that skinfold measurements should also be taken and obesity judged by this criterion. If the problem in these patients is in part due to excess deposition of fat, it would seem pertinent to monitor carefully the development of fatness in suspected patients. Criteria for the diagnosis of obesity have been suggested by Garn et al [1975, 1980] and standards for skinfolds and arm and muscle circumferences are available for clinical use [Frisancho, 1974]. If larger series of measurements and continued observations of the patients reported herein confirm these findings, measurements of skinfold thickness and upper arm circumference as well as MCPP analysis [Butler and Meaney, 1985] may provide a relatively simple method for aiding in the early diagnosis of infants suspected to have Prader-Labhart-Willi syndrome.

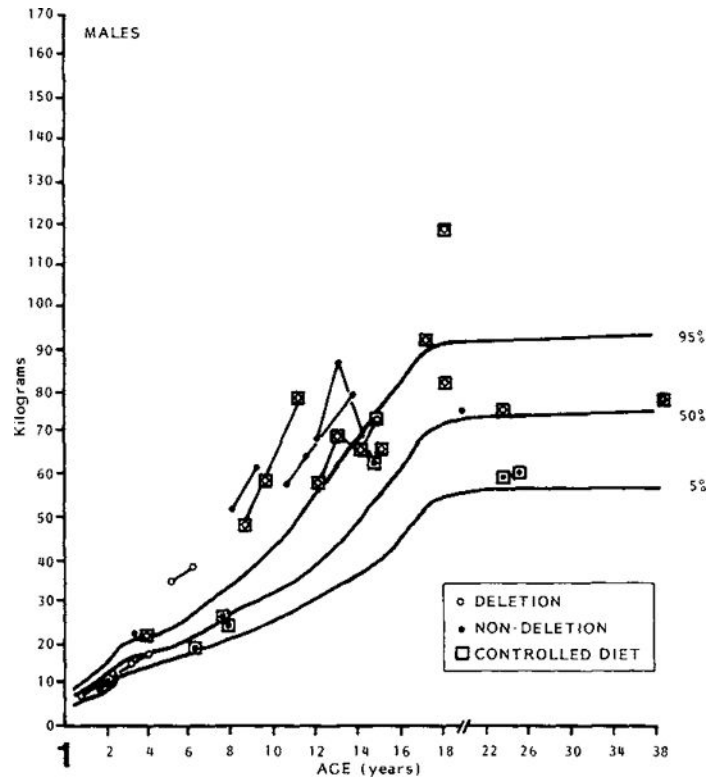
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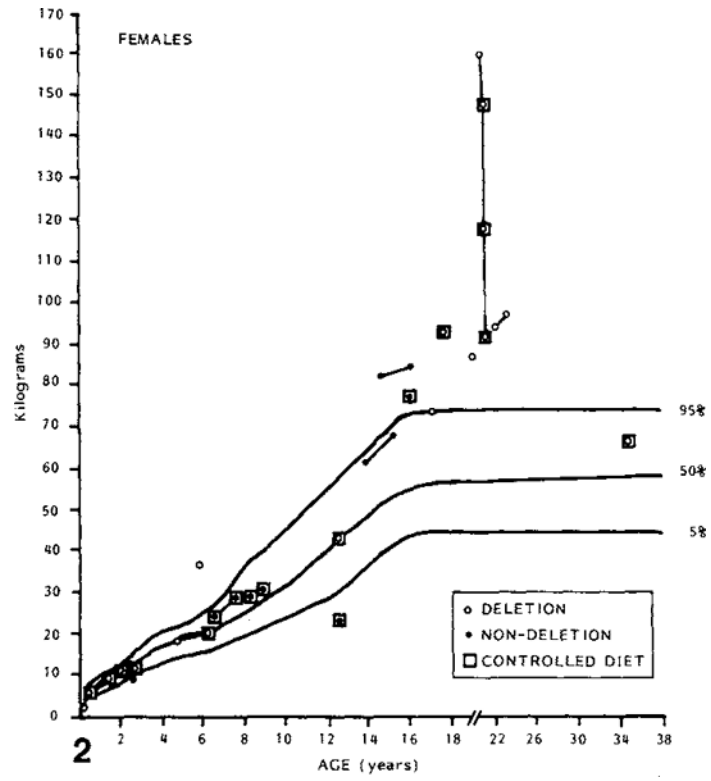
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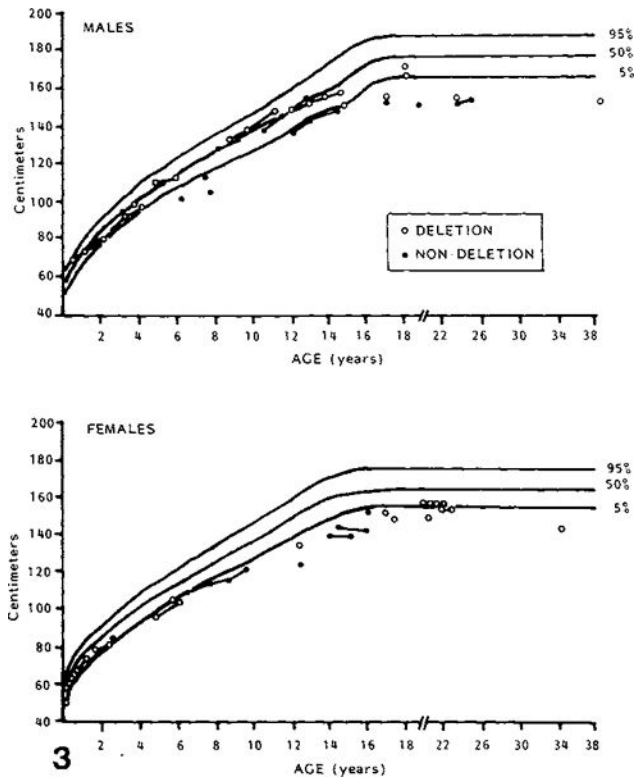
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**Fig. 1.**  
Weight of PLWS males plotted against chronological age.

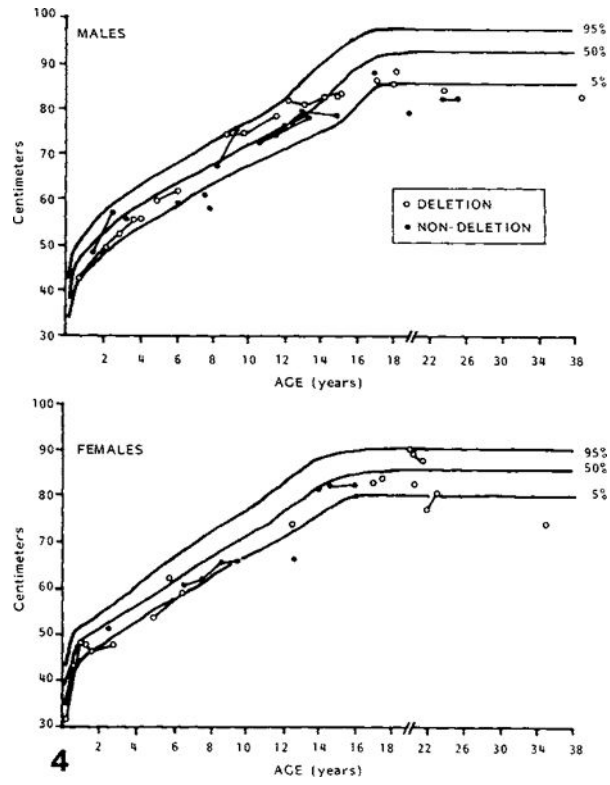


**Fig. 2.**  
Weight of PLWS females plotted against chronological age.

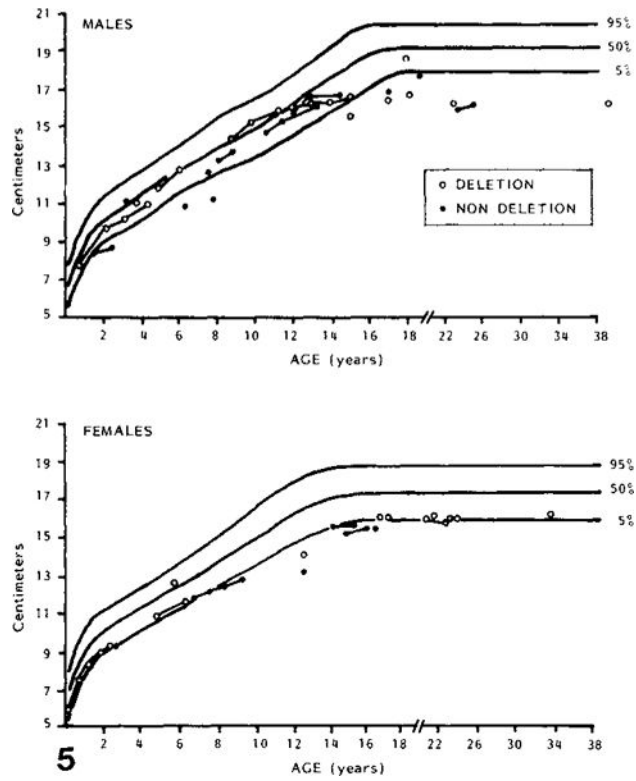


**Fig. 3.**  
Height of PLWS males and females plotted against chronological age.

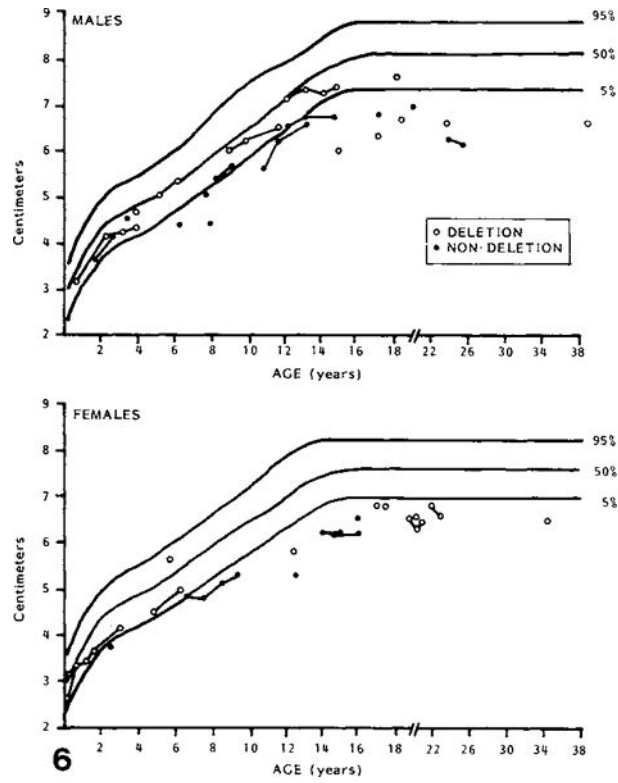




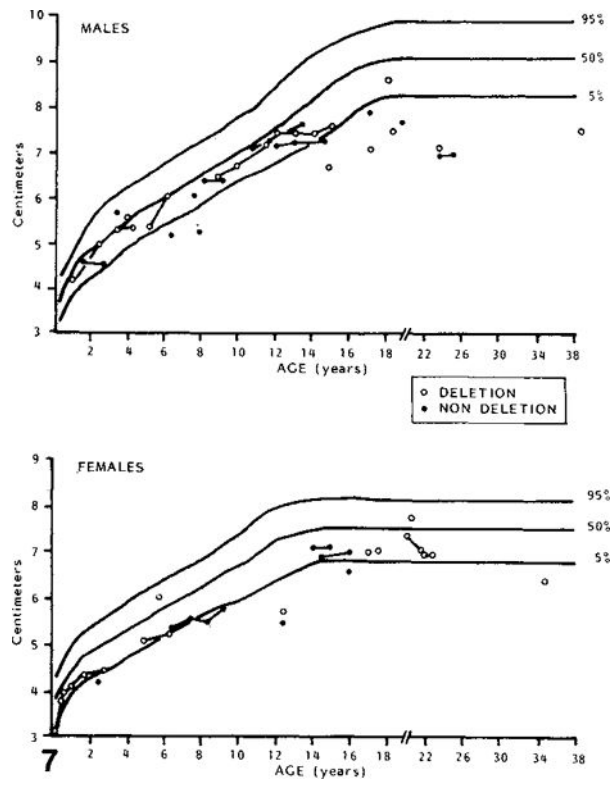
**Fig. 4.** Sitting height of PLWS males and females plotted against chronological age.



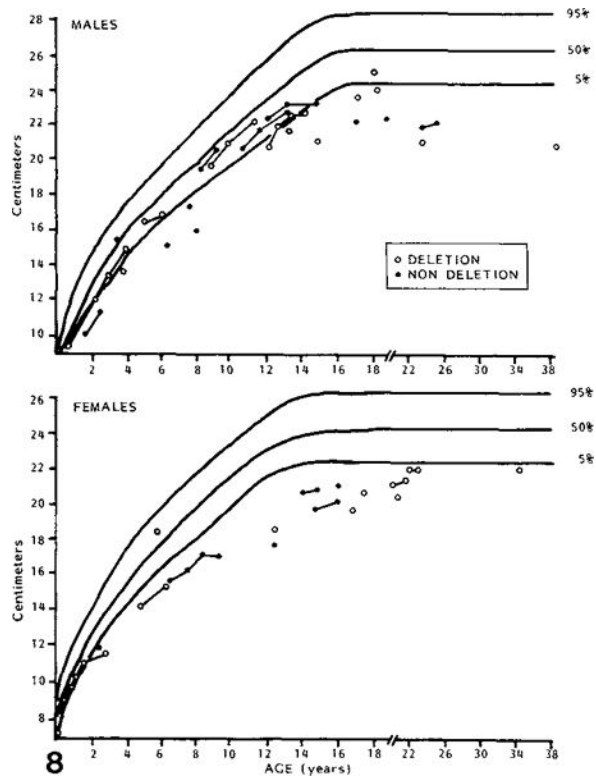
**Fig. 5.** Hand length of PLWS males and females plotted against chronological age.



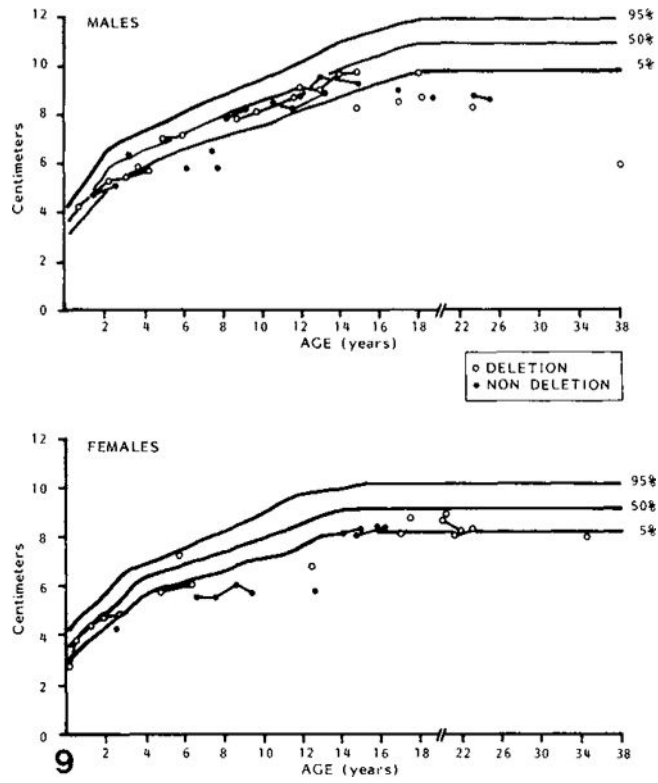
**Fig. 6.** Middle finger length of PLWS males and females plotted against chronological age.



**Fig. 7.**  
Hand breadth of PLWS males and females plotted against chronological age.



**Fig. 8.**  
Foot length of PLWS males and females plotted against chronological age.



**Fig. 9.** Foot breadth of PLWS males and females plotted against chronological age.

**TABLE I**  
Clinical and Cytogenetic Data From 38 Prader-Labhart-Willi Syndrome Individuals\*

Case No.	Sex	Birth date	Bone age (years)	Parental age (years)		BW (g)	BL (cm)	Karyotype	No. of times measured
				Maternal	Paternal				
1	F	9-23-81		23	25	2,210	48.0	del (15q)	10
2	M	7-27-80	BA=2 CA=2	30	31	2,980	48.3	del (15q)	5
3	M	10-28-78		22	21	4,170	53.3	del (15q)	1
4	F		BA = 6/12 CA = 10/12	23	25	2,550	48.3	del (15q)	3
5	M	10-19-76		30	39	2,580	53.3	del (15q)	2
6	F	1-7-77		26	27	2,690	52.1	del (15q)	1
7	M	10-23-72	BA = 10 CA = 10 1/2	21	23	2,550	48.3	del (15q)	3
8	F	5-10-70	BA = 13 1/2 CA = 13 1/12	28	28	2,300	45.7	del (15q)	1
9	M	7-19-69	BA = 11 1/2 CA = 11 10/12	20	35	2,410	53.3	del (15q)	4
10	M	11-23-67	Delayed	26	26	3,860		del (15q)	1
11	F	10-25-65	BA = 15 CA = 17 7/12	31	34	3,010	49.5	del (15q)	1
12	M	8-26-65		30	36	3,370	52.1	del (15q)	1
13	F	6-26-65	BA = 17 CA = 17	25	27	2,810	50.8	del (15q)	1
14	M	10-27-64		20	22	3,350	49.5	del (15q)	1
15	M	10-9-64	BA = 18 CA = 18 5/12	27	26	2,270	50.8	del (15q)	1
16	F	10-2-61		20	21	3,150	52.1	del (15q)	4
17	F	8-15-61		19	22	1,960	53.3	del (15q)	1
18	F	7-10-59		29	32			del (15q)	2
19	M	3-11-59	BA = 18 CA = 18	39	40	2,570	48.3	del (15q)	1
20	F	5-15-48		22	27	2,640	49.5	del (15q)	1
21	M	10-14-43				3,260		del (15q)	1
22	M	3-7-80		27	26	3,100	47.0	46,XY	2
23	F	9-25-79		21	23	2,310	48.3	46,XX	1
24	M	12-11-79		22	32	4,250	57.2	46,XY	1
25	M	10-22-76	BA=2 8/12 CA = 4	22	25	3,060	50.8	46,XY	1
26	F	10-14-74	BA=4 8/12 CA = 6 8/12	21	23	2,160	53.3	46,XX	4
27	M	6-21-75	BA = 5 4/12	31	34	3,520	48.3	46,XY	1

Case No.	Sex	Birth date	Bone age (years)	Parental age (years)		BW (g)	BL (cm)	Karyotype	No. of times measured
				Maternal	Paternal				
28	M	3-7-75	CA=5 7/12	24	24	2,610	48.3	46,XY	1
29	M	4-27-74	BA=7 9/12 CA=7 3/12	25	26	3,030	50.8	46,XY	3
30	M	11-4-70	BA=8	17	20	3,460	48.3	46,XY	3
31	M	7-22-69	CA=10 7/12 BA=10 1/2	26	24	3,200	50.8	46,XY	3
32	F	5-29-70	CA=11 10/12 BA=9	23	43	2,380		46,XX	1
33	F	8-16-67	CA=10 5/12 BA=8/12	37	47	3,350	48.0	46,XX	2
34	F	10-18-66	CA=1 2/12 BA=14 7/12	18	24	2,270	48.3	46,XX	2
35	F	11-22-66	CA=14 8/12 BA=15 10/12 CA=16 1/2	42	44	3,880	48.3	46,XX	1
36	M	5-25-65		21	22	3,000		46,XY	1
37	M	1-6-63		40	46	3,430	48.3	46,XY	1
38	M	11-4-57		19	21	2,840	52.1	46,XY	2

\* F, female; M, male; BA, bone age; CA, chronological age; BW, birth weight; BL, birth length.



**TABLE II**

Z Score Means and Standard Deviations for 15 Anthropometric Variables of Deletion and Nondeletion Prader-Labhart-Willi Syndrome Individuals

Variables	Deletion (N=21)		Nondeletion (N=17)	
	Mean	SD	Mean	SD
Weight	2.33	3.02	1.33	3.07
Height	-1.88	1.22	-2.43	1.42
Sitting height	-1.03	1.34	-1.43	1.25
Hand length	-1.60	1.12	-1.81	1.12
Finger length	-1.78	1.39	-2.41	1.14
Hand breadth	-1.73	1.63	-1.85	1.55
Foot length	-2.16	1.17	-2.34	1.32
Foot breadth	-1.70	1.70	-2.08	1.53
Head circumference	-1.18	1.06	-0.95	0.91
Head length	-0.86	1.06	-0.71	0.76
Head breadth	-1.72	0.85	-2.12	0.76
Arm circumference	2.47	2.08	1.91	2.70
Calf circumference	2.14	2.00	1.18	2.61
Triceps skinfold <sup>a</sup>	2.47	1.23	2.92	2.19
Subscapular skinfold <sup>a</sup>	2.88	1.92	3.85	3.93

<sup>a</sup>N= 15 for both subgroups.

TABLE III

Summary of Prader-Willi Syndrome Anthropometric Measurements

Measurement	Proband chromosome subgroup			
	Deletion		Nondeletion	
	n	%	n	%
Height				
<-2SD	9/21	(43)	9/17	(53)
<-1 SD	14/21	(67)	13/17	(76)
>1 SD	0/21	(0)	0/17	(0)
>2 SD	0/21	(0)	0/17	(0)
Weight				
<-2SD	1/21	(5)	2/17	(12)
<-1 SD	2/21	(10)	4/17	(23)
>1 SD	13/21	(62)	7/17	(41)
>2 SD	11/21	(52)	6/17	(35)
Sitting height				
<-2SD	4/21	(19)	4/17	(23)
<-1 SD	10/21	(48)	10/17	(59)
Upper arm circumference				
<-2SD	0/21	(0)	1/17	(6)
<-1 SD	1/21	(5)	2/17	(12)
>1 SD	16/21	(76)	8/17	(47)
>2 SD	11/21	(52)	7/17	(41)
Waist circumference				
<-2SD	0/18	(0)	0/17	(0)
<-1 SD	0/17	(0)	1/17	(6)
>1 SD	15/18	(83)	10/17	(59)
>2 SD	14/18	(78)	8/17	(47)
Calf circumference				
<-2SD	0/21	(0)	2/17	(12)
<-1 SD	0/21	(0)	4/17	(24)
>1 SD	15/21	(71)	6/17	(35)
>2 SD	11/21	(52)	6/17	(35)
Triceps skinfold				
<-2 SD	0/15	(0)	0/15	(0)
<-1 SD	0/15	(0)	0/15	(0)
>1 SD	14/15	(93)	12/15	(80)
>2 SD	9/15	(60)	10/15	(67)
Subscapular skinfold				
<-2 SD	0/15	(0)	1/15	(7)
<-1 SD	1/15	(7)	1/15	(7)
>1 SD	12/15	(80)	11/15	(73)

Measurement	Proband chromosome subgroup			
	Deletion		Nondeletion	
	n	%	n	%
>2 SD	10/15	(67)	11/15	(73)
Hand length				
<-2 SD	6/21	(29)	5/17	(29)
<-1 SD	14/21	(67)	13/17	(76)
Middle finger length				
<-2 SD	9/21	(43)	12/17	(71)
<-1 SD	14/21	(67)	14/17	(82)
Hand breadth				
<-2 SD	8/21	(38)	8/17	(47)
<-1 SD	14/21	(67)	10/17	(59)
>1 SD	0/21	(0)	1/17	(6)
>2 SD	0/21	(0)	0/17	(0)
Foot length				
<-2 SD	11/21	(52)	10/17	(59)
<-1 SD	19/21	(90)	13/17	(76)
Foot breadth				
<-2 SD	9/21	(43)	9/17	(53)
<-1 SD	13/21	(62)	11/17	(65)
>1 SD	1/21	(5)	0/17	(0)
>2 SD	0/21	(0)	0/17	(0)
Both hand and foot length				
<-2 SD	6/21	(29)	5/17	(29)
<-1 SD	14/21	(67)	13/17	(76)
Head circumference				
<-2 SD	6/21	(29)	2/17	(12)
<-1 SD	12/21	(57)	8/17	(47)
Head length				
<-2 SD	3/21	(14)	2/17	(12)
<-1 SD	10/21	(48)	7/17	(41)
Head breadth				
<-2 SD	9/21	(43)	9/17	(53)
<-1 SD	17/21	(81)	16/17	(94)
Frontal diameter				
<-2 SD	15/17	(88)	12/14	(86)
<-1 SD	17/17	(100)	14/14	(100)
Dolichocephaly				
C.I. <sup>a</sup> <75%	11/21	(52)	12/17	(71)
C.I. <sup>a</sup> <77%	14/21	(67)	15/17	(88)

<sup>a</sup>C.I. is cephalic index, which is equal to (Head breadth)/(Head length) × 100.