Hiroshima Journal of Medical Sciences
Vol. 31, No. 2, June, 1982
**HIJM** 31-12



# A Case of Aarskog Syndrome with a Review of Japanese Literature*)

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(Received February 19, 1982)

*Key words: Aarskog syndrome, Short stature, Shawl scrotum*

## ABSTRACT

A 9-month-old boy with Aarskog syndrome is described. Eleven cases of this syndrome were collected from the Japanese literature and reviewed. The most characteristic findings are short stature, craniofacial anomalies such as hypertelorism and broad nasal bridge, hand and foot anomalies, and shawl scrotum. If the pediatrician is familiar with these anomalies, especially short stature and shawl scrotum, more cases will be found.

## INTRODUCTION

Aarskog syndrome is characterized by short stature, peculiar facies, abnormalities of the hands and feet, and shawl scrotum. In 1970, this syndrome was first documented by Aarskog[1]. Since then about 50 cases have been reported in European and American[1,3,6,9,18,19]. Ohsaki et al.[16] reported the first case of this syndrome in Japan.

The purpose of this paper is to present a case we have recently studied and to review Japanese literature describing this syndrome. The case materials of the Aarskog syndrome were collected from Japanese literature during the eight-year period from 1974 to 1981. Twelve cases including ours were listed[2,8,12-14,16,20,21].

## CASE REPORT

This 9-month-old boy was born to a healthy mother after 38 weeks gestation with cephalic position. Pregnancy and delivery were uneventful. The birth weight was 3,033 g, length 48 cm, head circumference 32.5 cm and chest circumference 32 cm. The parents were unrelated.

Father and mother were 30 and 31 years old, respectively, at the time of birth. Father was 174 cm. Mother was short, 148 cm (−1.6 SD) and had widow's peak. There was no sibling.

At birth he was noted to have facial abnormalities, short hands, simian creases, and foot anomalies. However, the diagnosis was not made at this time. Early milestones were normal. Neither seizure nor neurological abnormalities were noted. At 9 months of age, his length was 68 cm, weight 6,880 g, head circumference 42.6 cm, and chest circumference 38 cm. Developmental quotient was 9 months by the method of Tsumori. His forehead was broad and prominent, and his hair line showed a pronounced widow's peak. The hypertelorism, antimongoloid slanting of the palpebral fissures, large cornea and ptosis were evident. The nose was short and stubby with anteverted nostrils, the nasal bridge was broad. His ears were low set. Hands and feet were short and broad. The 5th fingers showed clinodactyly and were short with a single ventral crease. The thin fingers showed hyperextensibility of the proximal interphalangeal and corpophalangeal joints and flexion of the distal interphalangeal joints. Each palm had a simian crease. The phallus

*) 相原克昭, 西 美和, 臼井朋包：Aarskog 症候群の一症例：本邦報告例の集計

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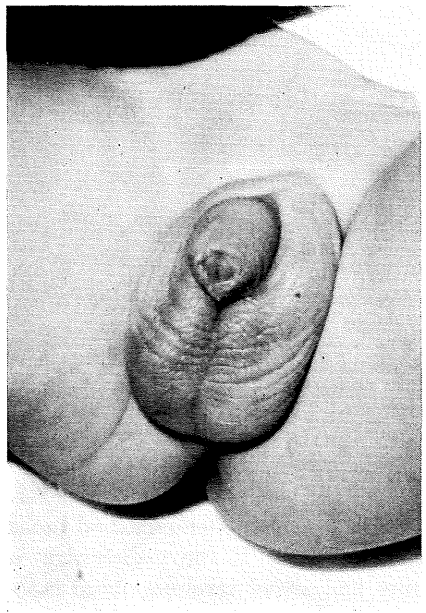

**Fig.** Scrotum surrounding phallus frontally: "shawl scrotum"

was normal. The scrotum was of normal size and the scrotal fold extended dorsally sorrounding the phallus "shawl scrotum" (Fig). Testes were palpable in the scrotum and of normal size.

Results of complete blood count and urinalysis were normal. Other laboratory data, including results of liver and renal function tests, serum electrolytes and thyroid function test, were all normal. Chromosome analysis was normal, 46, XY. Bone age was 6 months (Greulich and Pyle). Skull radiograph was normal. Spina bifida occulta of $Th_1$ through $Th_4$ was present in the vertebral X-ray film.

## DISCUSSION

Since Aarskog described a "new" syndrome in 7 male patients from 2 generations of the same family characterized by short stature, peculiar facies, abnormalities of the hands and feet, and a saddle abnormality of the scrotum[1], approximately 50 patients of about 20 families have been reported in European and American[1,3,4,6,7,9-11,15,17-19]. Our patient showed typical features of the Aarskog syndrome. In Japan, since the first case of this syndrome was documented in 1974[16], twelve cases including ours have been reported[2,8,12-14,16,20,21]. Clinical findings of these 12 patients were summarized in Table as compased with the reports of Berman et al.[3] and Escobar et al[6].

Short stature was one of the most characteristic findings, which was present in 100% in both Japanese and European and American patients. Craniofacial anomalies were also characteristic findings in Japanese patients and European and American patients; hypertelorism was noted in 92% in the former and in 87% in the latter, broad nasal bridge in 100% in the former and 87% in the latter, and short nose in 100% in the former and 95% in the latter. Anomalies of hands and feet were also observed frequently as shown in Table. A characteristic genitalia with scrotum fold overriding the base of the penis "shawl scrotum" was observed in 100% in Japanese and in 95% in European and American patients.

The mode of inheritance of this syndrome is likely to be X-linked recessive or autosomal dominant inheritance with sex-limited expression[1,3,6,7,9,19]. Carrier females often show several minor abnormalities[3,6,9,19], as was the case in the mother of our patient. She had short stature and widow's peak. This syndrome may not be as rare in Japan as may appear recently from reports in literature.

The problem for the pediatrician today is familiarization with the ever increasing number of new syndromes. It is necessary to find "shawl scrotum" in a short child with peculiar facies and abnormalities of the hands and feet for making a diagnosis of Aarskog syndrome, since "shawl scrotum" is seldam noted in the other syndromes.

Short stature is a consistent feature of this syndrome, however, the cause of this growth retardation is unknown. Plasma growth hormone(GH) concentrations have been found to be normal[1,18] although detailed endocrinological documentaion is lacking in almost all patients. Aarskog found no growth response to treatment with human GH in only one case[1]. In Japanese patients, GH secretion was almost normal[2,13,14,16,21], but one case showed low response to insulin and arginine loading test[2], and another had isolated GH deficiency[12]. Although these two unknown entities of Aarskog syndrome and GH deficiency might be coincidental, a complete endocrinological study is necessary in Aarskog syndrome whose growth curve deviates progressively from the third percentile.

**Table.** Clinical findings in 12 Japanese Aarskog syndrome

| Case | 1 | 2 | 3 | 4 | 5 | 6 | 7 | 8 | 9 | 10 | 11 | 12 | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| Author | Ohaski | Fujisawa | Murata | Yamauchi | Nagashima | | Tamura | Abe | | | Kodama | present case | total | Berman & Escobar |
| Date | 1974 | 1977 | 1978 | 1980 | 1980 | | 1980 | 1981 | | | 1981 | 1982 | (%) | 1975 & 1978 |
| Short stature | + | + | | + | + | + | + | + | + | + | + | + | 11/11(100) | 39/39(100) |
| Craniofacial | | | | | | | | | | | | | | |
| Hypertelorism | + | + | + | + | + | + | + | + | + | + | − | + | 11/12( 92) | 34/39( 87) |
| Widow's peak | | ± | | − | + | + | + | + | − | + | + | + | 7/10( 70) | 18/39( 46) |
| Ptosis | | + | + | + | + | − | + | − | − | − | − | + | 6/11( 55) | 23/39( 59) |
| Antimongoloid slant | + | + | | + | + | + | + | − | + | − | + | + | 9/11( 82) | 18/39( 46) |
| Maxillary hypoplasia | | + | | − | | | − | + | + | + | − | − | 4/ 8( 50) | 6/39( 15) |
| Broad nasal bridge | + | + | + | + | + | + | + | + | + | + | + | + | 12/12(100) | 34/39( 87) |
| Short nose | | | + | + | + | + | + | + | + | + | + | + | 10/10(100) | 37/39( 95) |
| Long philtrum | + | + | | + | | | + | + | + | + | + | − | 8/ 9( 89) | 32/33( 97) |
| Ear anomalies | | + | + | + | + | ? | − | | | | + | + | 6/ 7( 86) | 22/39( 56) |
| Hands and feet | | | | | | | | | | | | | | |
| Foot anomalies | | + | | − | + | ? | + | + | + | − | + | + | 7/ 9( 78) | 20/39( 51) |
| Short fingers | + | + | | + | + | + | + | + | + | + | + | + | 11/11(100) | 35/39( 90) |
| Short & broad hands | + | + | | | + | + | + | | | | | + | 6/ 6(100) | 35/39( 90) |
| Simian creases | | + | | + | + | + | + | + | + | − | − | + | 8/10( 80) | 14/26( 54) |
| Interdigital webbing | + | + | | + | + | + | | + | | | | − | 6/ 7( 86) | 16/39( 41) |
| Genitalia | | | | | | | | | | | | | | |
| Shawl scrotum | + | + | + | + | + | ? | + | + | + | + | + | + | 11/11(100) | 37/39( 95) |
| Cryptorchism | + | + | + | + | + | + | − | + | + | + | + | − | 10/12( 83) | 29/39( 74) |
| Inguinal hernia | | − | | − | + | + | − | + | + | + | − | − | 5/10( 50) | 23/39( 59) |
| Others | | | | | | | | | | | | | | |
| Mental retardation (mild) | − | | | − | − | − | − | + | − | + | + | − | 3/10( 30) | 5/39( 13) |
| Spinal anomalies | | | | − | − | ? | | − | + | − | | + | 2/ 6( 33) | 13/39( 33) |

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