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Weill-Marchesani Syndrome

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Weill-Marchesani syndrome is a rare systemic connective tissue disease characterized by short stature, brachydactyly, ectopia lentis and spherophakia. This was first reported by Weill in 1932 and subsequently well characterized by Marchesani in 1939(1). Genetically this syndrome often shows autosomal recessive inheritance. Frequent consanguinity between parents support this model. There have been reports suggesting autosomal dominant inheritance(2).

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Gorlin *et al.* reported a family in which a father and two of his three children were affected suggesting genetic heterogenecity or the possibility of pseudodominance(l).

We report six patients with Weill-Marchesani syndrome from 4 sibships, giving an account of family data to help to .delineate the mode of inheritance. This is the largest and the first report from India.

Case Reports

The important features noted in our six cases are summarized in *Table I.*

Discussion

In 1932 during research on Marfan syndrome, Weill noted that of the 8 individuals he was studying with presumed Marfan syndrome one was short in stature and had short swollen fingers with limited range of motion(l). Later, in 1939 Marchesani described the association of spherical lenses and brachydactyly in two families and suggested the term of dystrophia mesodermalis hyperplastica which is presently designated as Weill-Marchesani syndrome to distinguish from those with dystrophia mesoder-

TABLE I–*Summary of Cases*

	Family I		Family II		Family III	Family IV
	Patient 1	Patient 2	Patient 3	Patient 4	Patient 5	Patient 6
Age	9	50	25	35	5	12
Sex	Male	Male	Female	Male	Male	Male
Consanguinity	+	-	+	-	+	+
Family history	+ (Son of Pt. 2)	+ (Father of Pt. 1)	+ (Sister of Pt. 4)	- (Brother Pt. 3)	-	-
Eyes						
Loss of vision bilateral	+	+	Defective vision	Defective vision	Defective vision	Defective vision
Age of onset (yrs)	8	10	15	15	4	10
Nystgmus	+	-	-	-	-	-
Squint	+	-	-	-	-	-
Hazy cornea	+	-	+	-	-	-
Iridodonesis	+	-	-	-	-	-
Phakodonesis	_	-	-	-	-	-
Subluxation of Lens	-	-	+ (Lt)	+(Bil)	+(Bil)	-
Dislocation of Lens			+(Rt)	-	-	-
Spherophakia	-	-	-	-	-	+
Myopia	+	-	+	+	+	+
Hands & Feet						
Short Flat feet	-	+	-	-	+	+
Brachydactly	-	-	+	+	-	-
Short stiff fingers	+	+	-	-	+	-
Others						
Short stature	+	+	+	-	-	+
Other systems	Normal	Normal	Normal	Normal	Normal	Normal

malis hypoplastica in the Marfan syndrome. Subsequent observers emphasized the dichotomy in musculoskeletal features of Weill-Marchesani syndrome and Marfan(2).

Ectopia lentis is a common manifestation of 3 heritable disorders namely Marfan syndrome, homocystinuria and Weill Marchesani syndrome. The musculoskeletal manifestations of Weill-

Marchesani and Marfan syndromes present sharp contrast. A typical Marfan syndrome patient is tall, lean, with high arm span, arachnodactyly and hyperextensible joints(l). While Weill-Marchesani syndrome individuals are short in stature, with brachymelia, brachydactyly, stubby spade-like hands and feet, and limitation of mobility of joints. Two of our patients had limitation of movements of joints.

A variety of manifestations of Weill-Marchesani syndrome can present a diagnostic problem. A diagnosis should be based only on the combination of specific ocular and skeletal abnormalities. Isolated ocular or skeletal features can occur as isolated familial anomaly or as a part of other syndromes(2).

The important anomalies observed in this syndrome are of ocular origin. They include spherophakia, iridodonesis, ectopia lentis, lenticular myopia and pigmentary degeneration of fundii, optic atrophy and glaucoma(2,4). Glaucoma, which frequently occurs, leads on to blindness in many cases.' Glaucoma can result either due to subluxation of lens or even without subluxation. The spherophakia produces shallow anterior chamber thus producing angle narrowing(4). These two mechanisms in the production of glaucoma in Weill-Marchesani syndrome are evident, also among our patients where one patient had both subluxation of lens and glaucoma (Family I, Case 1), while another had developed glaucoma even without subluxation of lens (Family IV, Case 1). To maintain vision, an early diagnosis is necessary. Peripheral iridectomy, lens extraction and recently laser iridotomy are used in the treatment(5).

The precise mode of inheritance of Weill Marchesani syndrome is still uncertain. Most of the reports in literature support an autostomal recessive pattern of inheritance(l). The heterozygous patients may have short stature or brachydactyly and may suffer from refractive errors but not ectopia lentis. The consanguinity among parents of all our cases also suggests autosomal recessive inheritance. In the present report in one family father and his son were affected *(Fig. 1),* giving an impression of an autosomal dominant inheritance pattern. However, a high degree of inbreeding in that family pedigree *(Fig. 2)* makes it difficult to decide the type of inheritance. The death of siblings and uncles of the

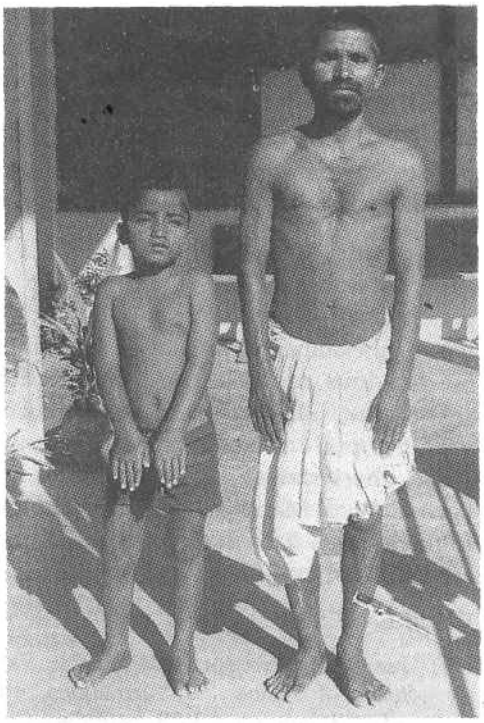

Fig. 1. Photographs of patients 1 and 2 showing short stature and brachydactyly.

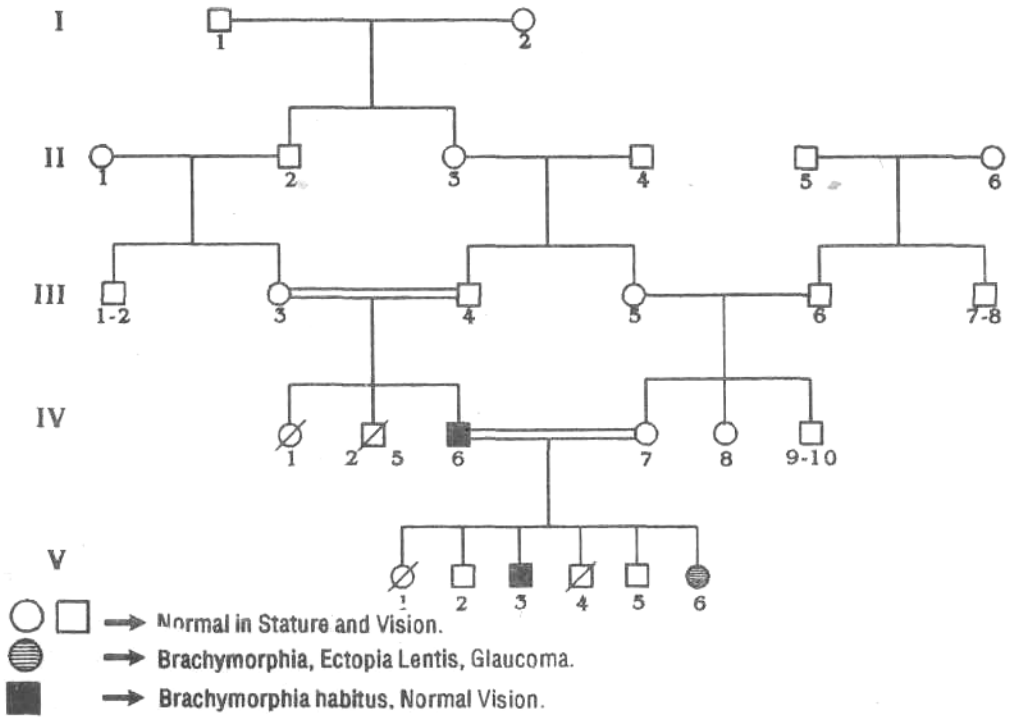

Fig. 2. *Pedigree of Family 1*

proband were unrelated to the present problem.

Recently, Verloes *et al.* described a Weill Marchesani like syndrome in 3 generations of one family showing dominant inheritance^) and reviewed similar previous reports. They proposed a new name, GEMSS syndrome (glaucoma, ectopia, microspherophakia, stiff joints, short stature) for dominantly inherited Weill Marchesani like syndrome to distinguish it from the classical recessive Weill Marchesani syndrome(6).

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