WEILL-MARCHESANI'S SYNDROME: FAMILIAL INVOLVEMENT

SÍNDROME DE WEILL-MARCHESANI: AFECTACIÓN FAMILIAR

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ABSTRACT

Case report: We report the case of a child short in stature with brachydactyly and brachymorphy who was referred to our office complaining of poor vision. This was a case of Weill-Marchesani’s syndrome described in a family, in which four of the eight children were affected by spherophakia, brachymorphy and brachydactyly.

Discussion: There are few familial cases of Weill-Marchesani’s syndrome reported in the literature. Both autosomal dominant and recessive inheritances have been described. The ophthalmologist plays a crucial role in its diagnosis and management, since the ocular involvement is the most severe one (Arch Soc Esp Oftalmol 2006; 81: 349-352).

Key words: Weill-Marchesani’s syndrome, familial involvement, ectopia lentis, spherophakia.

INTRODUCTION

The Weill-Marchesani syndrome is an infrequent alteration of the connective tissue which involves the eyes. Patients exhibit low height, brachydactyly with joint stiffness, microspherophakia, progressive lens myopia, lens luxation and secondary glaucoma. Since the syndrome was described by Weill and Marchesani in 1932 and 1939 (1), researchers have documented patterns of dominant and recessive autosomic inheritance (1-3).

RESUMEN

Caso clínico: Presentamos el caso clínico de un niño con talla baja, braquidactilia y braquimorfia remitido a la consulta por baja visión. Se trata de un caso de síndrome de Weill-Marchesani en una familia en la que cuatro de los ocho hijos presentan esferofaquia, braquimorfia y braquidactilia.


Palabras clave: Síndrome de Weill-Marchesani, afectación familiar, ectopia lentis, esferofaquia.
CASE REPORT

A 13-year old boy is referred for ophthalmological assessment due to poor eyesight. At the phenotype level, the boy was short (1.33 m tall), with brachymorphy and brachydactyly in all limbs (figs. 1 and 2).

The patient exhibited a corrected refraction error of −13.00–6.50 to 1° in the right eye and of −16.00–6.25 to 179° in the left one. With this correction, his visual acuity reached 0.4 and 0.2 respectively. He did not exhibit monocular dyopia or findings in extrinsic or intrinsic eye motility.

The horizontal corneal diameter was of 12.0 mm in both eyes, and pachymetry was of 613 and 611 microns respectively. The anterior chamber was narrow and bilateral iridophacodonesis was observed. Microspherophakia was evidenced, with anterior displacement of both lenses within the posterior chamber (fig. 3).

The patient’s IOP was of 20 mmHg bilaterally. The gonioscope exploration revealed a symmetrical small angle in both eyes, rated 2 in Schaffer’s scale.

Evaluation with Orbscan-II topograph (corneal topograph, Bausch and Lomb, U.S.A.) and an eye fundus exploration did not reveal alterations.

To prevent closure of the angle, a Nd:YAG laser iridotomy was carried out in both eyes, at 10:00 and 2:00 respectively. Due to vision stability and absence of dyopia, a conservative approach was adopted according to the patient’s wishes. The family was checked for visual acuity, IOP, photo documentation and visual field explorations to detect the appearance of glaucomatous damages.

A review of the patient’s family history revealed that he was the younger child in a family of eight siblings, in which - in addition to the subject - one brother and two sisters exhibited spherophakia, brachymorphy and brachydactyly (fig. 4). Both parents were healthy and none of their children has had descendants.

DISCUSSION

The clinical diagnosis of the Weill-Marchesani syndrome is based on lens and skeletal abnormalities.

In addition to small height, brachydactyly and brachymorphy, ocular alterations can be found in the patient.
such as lens myopia, microspherophakia, ectopia lentis and secondary glaucoma (2). At present, it is considered that spherophakia is not a requirement for the diagnostic (1). Other less frequent findings include vitreous liquefying, choriorretinal degeneration, megalocornea and microcornea (1).

A normal lens is nearly spherical at birth, with an equatorial diameter of 6.5 mm and anteroposterior (sagittal) diameter of 3.5 to 4.0 mm. The equatorial diameter grows rapidly to reach 9.0 mm at age 15, and maintains this size permanently. In contrast, the sagittal diameter is of 3.7 mm at age 20 and 4.0 at age 50, reaching 4.75-5.0 mm at age 80-90. Most of the physiological changes of the lens occur in the anterior surface, which becomes more spherical with aging (2). In microspherophakia, the lens is small and spherical. The cause of microspherophakia is unknown although it has been suggested that it is due to an alteration in the development of secondary lens fibers and/or a defect in the insertion of abnormally thin secondary fibers. Other theories attribute microspherophakia to the lack of tension in the rudimentary zonular fibers which inhibits the physiological development of the lens and leads to the spherical shape. The sagittal diameter of the normal lens changes very little during childhood and puberty because an adequate tension in the zonular fibers limits expansion along the sagittal plane. An abnormality in the zonular fibers would allow the lens to remain spherical instead of tending to a bi-convex shape with the passage of time (1).

The above, linked to the tendency towards dislocation and displacement of the lens, would explain the progression of lenticular myopia.

Significant myopia appears at the average age of 11.2 whereas the subluxation of the lens appears at the age of 18.2 and its displacement usually takes place inferiorly, contrary to Marfan’s syndrome and homocystinuria (2). In our 13-year old patient we observed myopia and lens ectopy, although the lens dislocation occurred in the anterior direction.

The loss of vision is due to myopia and to the glaucomatous damage caused by the progressive narrowing of the anterior chamber and closure of the angle (2).

Indications for lens surgery are dislocation to the anterior chamber, progressive subluxation with pupil bisection, imminent dislocation, monocular diplopia and visual acuity below 20/70 (far sight) and J5 for near sight (4).

Present-day management comprises lens extraction with or without placement of intraocular lens and anterior vitrectomy (4), in addition to medical or surgical treatment of the secondary glaucoma. However, there is a greater risk of postop complications in these patients, such as athalamia after filtrating surgery (5) or zonular ruptures (2,5).

The assumed disparity between the Weill-Marchesani syndrome and dominant/recessive inheritance patterns is not yet clear. Traditionally, lens ectopy was considered to be a recessive autosomic trait, although there are documented cases of dominant autosomic inheritance. The English-language acronym GEMSS (glaucoma, ectopia lentis, microspherophakia, stiff joints, small height) has been suggested to describe the dominant form, which exhibits a possible relationship with the Moore-Federman syndrome, with lens ectopy being the differentiating characteristic between both entities (3). The scarcity of documentation about families affected by these syndromes calls for a greater number of contributions in order to elucidate their transmission. The Spanish literature which we consulted (Medline) did not provide references to any family having more than one member affected by the Weill-Marchesani syndrome.

As mentioned above, the gene could be dominant with reduced penetration and a broad range of expressions due to the finding of small height, brachydactyly and spherophakia in several members of the family. Another alternative would be the

Fig. 4: Members of the subject’s family, wherein the three brothers at the patient’s right are affected.
incomplete recessive nature of the gene, with partial expression in heterozygotes (1,3). Considering the small height of the father, pseudo-dominance as inheritance pattern cannot be excluded.

Wirtz has suggested that 15q21.1. would be the gene which accounts for the Weill-Marchesani syndrome (3). The ophthalmologist carries out an essential diagnostic and prognostic work in systemic pathologies involving the eyesight, such as the Weill-Marchesani syndrome. Early optical correction prevents amblyopy and adequate follow-up of patients allow the ophthalmologist to detect and treat secondary glaucoma.

REFERENCES