Case Reports:

Primary congenital glaucoma in mother and her son

Abstract:
Primary congenital glaucoma (PCG) is a rare eye disease that affected 0.01-0.04% of total blindness. The disease is usually manifested at birth or before 3 years of age. The incidence of PCG is different in different populations and is approximately 1 in 10,000 births in western developed countries. The diagnosis by the age of 6 months is about 60%, and 80% within the first year of life... Males is common (about 65%), and the involvement is usually bilateral is about 70%. The disease is family in 10-40% of cases with variable penetrance (40-100%). Patients with a familial pattern usually show a recessive pattern with incomplete or variable penetrance and possibly multifactorial inheritance. PCG relates an isolated maldevelopment of the trabecular that can raise the IOP. PCG is the most common form of developmental glaucoma. The condition is typically bilateral and 30% of the cases may be unilateral. Surgical treatment with trabeculotomy and medical therapy is a supportive role. The prognosis for children with PCG is quite variable, with some achieving good vision thanks to scientific advances, while others go blind especially in a developing country. So the quality of life in these patients is very necessary. Our 2 cases of PCG on a mother and her son in one family are reported with some satisfying results in the quality of life.
Key words: Primary congenital glaucoma, surgical treatment, medical therapy, quality of life.

1. Introduction:

“There are 3 glaucoma patents relating to age of onset: Congenital glaucoma: The glaucoma exists at birth, and usually before birth. Infantile glaucoma: Occurs from birth until 3 years of life. Juvenile glaucoma: Occurs after the age of 3 to teenage years. Primary congenital glaucoma (PCG) is a rare eye disease that affected 0.01-0.04% of total blindness. The disease is usually manifested at birth or before 3 years of age. The incidence of PCG is different in different populations and is approximately 1 in 10,000 births in western developed countries” [1].
“The diagnosis by the age of 6 months is about 60%, and 80% within the first year of life. Male is common (about 65%), and the involvement is usually bilateral is about 70%. The familial disease is in 10-40% of cases with variable penetrance (40-100%). Patients with a familial pattern usually show a recessive pattern with incomplete or variable penetrance and possibly multifactorial inheritance. An autosomal dominant and pseudo-dominant mode of inheritance has also been reported” [2][3].
“Genetic studies: Loci of recessively inherited PCG (gene GLC3) have been identified by genetic linkage analysis” [4]. The majority of congenital glaucoma map to the GLC3A locus on chromosome 2 (2p21). Families linked to these loci display severe phenotypes with autosomal recessive inheritance patterns. Glaucoma with autosomal dominant inheritance pattern has been shown to chromosome 1q23-q25 (TIGR/MYOC gene).
PCG relates an isolated maldevelopment of the trabecular that can raise the IOP. PCG is the most common form of developmental glaucoma. The condition is typically bilateral and 30% of the cases may be unilateral.
Management; Surgical treatment included Primary surgical treatment is usually with goniotomy or trabeculotomy, combined trabeculotomy. Medical therapy is a supportive role. The prognosis for children with PCG is quite variable, with some achieving good vision thanks to scientific advances, while others go blind especially in a developing country. So the quality of life in these

patients is very necessary [5]. In this paper 2 cases of PCG on a mother and her son in one family are reported with some satisfying results in the quality of life.

Case Reports:

Two cases of primary congenital glaucoma PCG on a mother and her son in one family are reported. (Figure attach)

Case 1: Microphthalmia/PCG. A 45 –year old female with microphthalmia in the right eye and left eye with PCG are presented. She is a housewife. He is checked up his health by the Charitable Team of Health Care Examination period yearly.

General state: Height= 1, 55 meter; Weight=53 kg; Pulse = 75/mn; Arterial tension=120/75 mmHg. Mental =Development normal. Nothing abnormal detecting (N/A).Ophthalmology examination: Right eye (RE) = Microphthalmia at birth. Left eye (LE) = cornea opacity. The anterior segment is not exam because opacity of cornea. Visual acuity (VA) of LE is Light perception (+). Intraocular pressure (IOP) of LE = 30mmHg.

LE=PCG. She was diagnosis with LE=PCG. Use acetazolamide (oral) and pilocarpine (drop). She did not receive any surgical treatment. VA /IOL was not restored.

Case 2: A 12 -year old boy with a PCG in both eyes. He was the son of the case1 and they are living together. His mind is well developed. He has also checked up on his health with his mother. General state: Height= 1, 3 meter; Weight=35 kg; Pulse = 70/mn; Blood pressure=110/65 mmHg. Nothing abnormal was detected (N/A). Ophthalmology examination: RE with trabeculectomy= Deep anterior chamber, mild iris change. Pupil 4mm diameter was not responsible for the light. LE = Deep anterior chamber with the opacity of the crystalline lens.VA of both eyes (OU) are blind, no perception of light. IOP of both eyes is 30mmHg.

Treatment: RE=Trabeculectomy when he was 7 years old with VA 1/10 and 5 years later trabeculotomy 5 years ago VA was no perception of light and IOP was not restored.

Discussion: Management of PCG included surgical treatment and medical therapy. Surgical treatment: Primary surgical treatment is usually with goniotomy or trabeculotomy, combined trabeculotomy.

Medical therapy: Medical therapy usually plays a supportive role. In one study, in 161 eyes with congenital glaucoma: Medical therapy alone reduced the IOP to less than 21 mm Hg in 12% of eyes in the short term and 10% of eyes in the long term. There are some drugs [6]:

1. Beta-blockers:
2. Carbonic anhydrase inhibitors (CAIs)
3. Prostaglandin analogs
4. Alpha-2 agonists
5. Other drugs pilocarpine, mannitol

Management of Residual Vision in Pediatric Glaucoma

With the best treatment, many children with PCG having ended up result with “low vision”. Visual rehabilitation and low vision aides can help the children tor a normal or near-normal life with near-vision. The use of these devices should have cooperated with the child and parents [7].

The earlier diagnosis can be confirmed, the more likely it can be treated effectively. The prognosis of PCG is quite variable, with some achieving good vision thanks to scientific advances. Do hope that ongoing research in this field will assist to further help the preservation or restoration of vision of children suffering from glaucoma. A lot of children suffering from glaucoma will go blind especially in a developing country. So the quality of life in these patients is very necessary.

Palliative care: “The role of palliative care is to relieve the suffering of patients and their families

by the comprehensive assessment and treatment of physical, psychosocial, and spiritual symptoms patients experience" [8].

Quality of life: Our two patients are continuing to live, to participate in all activities with the community with volunteers / donate from people around the world. When she was born she had been sent to a "house of love" located in or near a pagoda that was organized by non-government organization NGO, Church (Catholic), Pagoda (Buddha). When she grew up she became one of many volunteers of this house. Up to 27 years old she had married and 6 years later she was born her son. The period of follow-up of our 2 patients was between 12-45 years.

"All of these as a part of palliative care can be a reason to make the patients decreasing worry about their disability. Despite a developing country the quality of life index of Vietnam is ranged from 60 in 2018" [9].

Conclusion

Thanks to scientific advances but PCG finally vision is blind especially in developing countries. So the quality of life in these patients played an important role by a non-government organization. This means they can live can participate with the community as well as decreasing worry about their disability. Do hope ongoing research in this field will assist to further help the preservation or restoration of vision of children suffering from primary congenital glaucoma.

Consent

As per international standard or university standard, patient(s) written consent has been collected and preserved by the author(s).

Ethical Approval:

As per international standard or university standard written ethical approval has been collected and preserved by the author(s).

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Conflict of Interest: No financial disclosures

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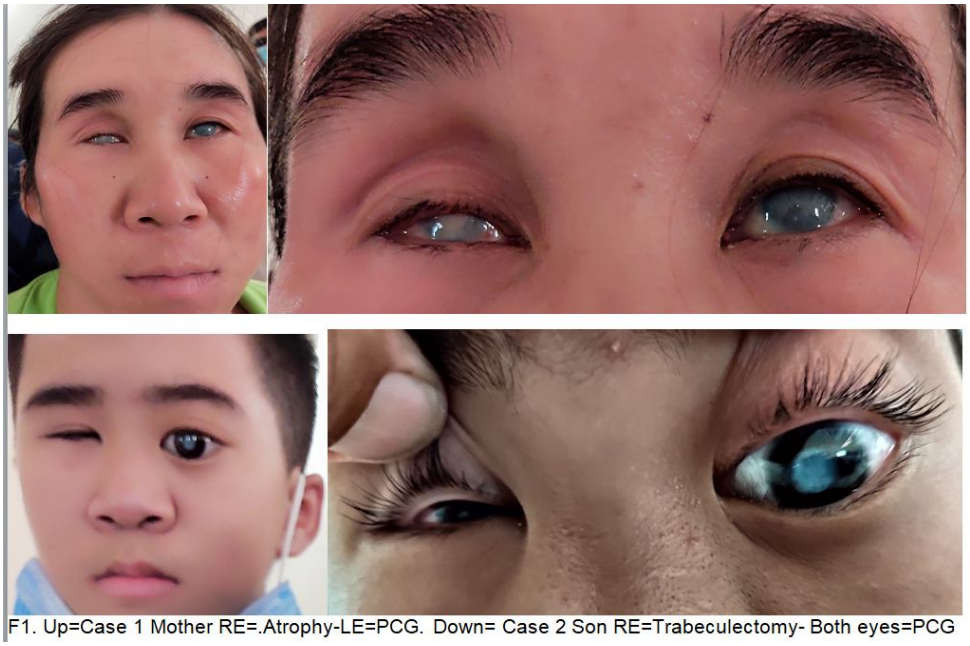

F1. Up=Case 1 Mother RE=.Atrophy-LE=PCG. Down= Case 2 Son RE=Trabeculectomy- Both eyes=PCG