A CASE OF GOLDENHAR SYNDROME ASSOCIATED WITH AN INGUINAL HERNIA AND EXTENSIVE RENAL, URETERIC, AND BLADDER ABNORMALITIES

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Goldenhar's syndrome, oculo auriculo-vertebal dysplasia is a congenital anomaly involving mainly the eyes, ears, facial bones and vertabrae that is, mainly those structures arising from the first and second branchial arches, the first pharyngeal pouch, the first branchial cleft and the primordia of the temporal bone. Its frequency among males and females is approximately the same.

The most consistent manifestations of the syndrome are epibulbar dermoids and lipodermoids, preauricular appendages and coloboma of the upper lid. (Baum & Feingold 1970).

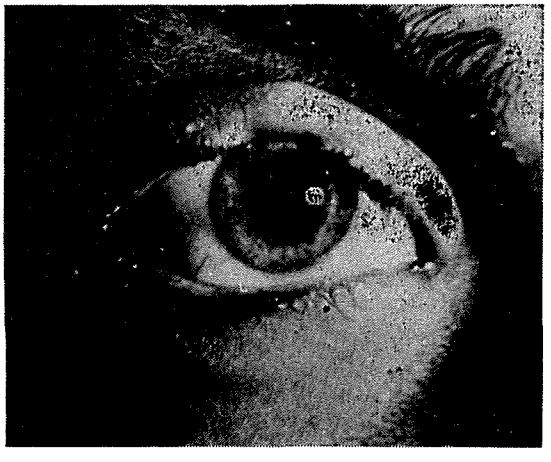
However, systemic abnormalities have been reported such as cardiac abnormalities, mental retardation, hydrocephalus and renal abnormalities (GROSS 1905). It has not been possible to find a common etiological explanation for all the manifestations of this syndrome. In the majority of cases, the defects do not seem to be hereditary (Baum & Feingold 1970).

The case under discussion, now 17 years old, shows besides the most consistent manifestations that is coloboma of the left upper lid, a large bulbous lipodermoid at the outer canthus right eye and numerous preauricular appendages, in front of the right of left auricles, shows various abdominal abnormalities in con-nexion with the kidneys, ureters and testicles.

He was first seen at the out-patients department, St. Luke's Hospital when 4½ years old, suffering from a distended abdomen.

He was born in January 1960 and his weight was 8st 8lbs. He has a 10 year old sister completely normal. Coloboma of the left upper lid, numerous preapricular appendages and a fairly large dermolipoma outer, canthus right eyes were also observed.

The coloboma Fig 1. is central,



quardrangular in shape, about half an inch long and a quarter of an inch wide. It leaves uncovered the upper part of the cornea. This is the first case ever seen in Malta.

On adduction of the right eye, there comes into view a large multilobar dermo-lipoma with numerous eye lashes. The latter require periodical removal.

There are three scars in front of the left Tragus and two scars in front of the right one. The auricular appendages were removed when the patient was four years old.

In September 1964, at the insistence of Prof, A Craig, he was transferred to the paediatric urological unit at the Hospital for Sick Children. Great Ormond street, as suffering from congenital bilateral Hydronephrosis and possibly a bladder neck obstruction. A radiological report showed a double kidney on the right and a hydronephrosis of the left kidney. An intravenous pyelogram revealed "a moderately hyronephrotic right kidney and a grossly hydronephrotic left kidney which appeared to be displaced downward and outwards. A micturating aystogram revealed a huge filling defect on the left side of the bladder.

He was operated upon for a left ectopic ureterocele on 16-10-64. The left kidney was explored on 16-10-64. The left kidney was explored. A huge hydronephrotic upper pole was found with an enormous upper pole ureter. The lower pole was also hydronephrotic, but some reasonable ammount of renal tissue remained.

An upper pole hemi-nephroureterectomy was therefore performed and the lower end of the upper pole ureter was exteriorised on the skin.

He was operated upon 5 days later. The reminder of the upper pole ureter of the left kidney was removed and the huge ureterocele associated with this was decapped from within the bladder.

The present situation therefore is that this litlle boy has a duplex kidney on the right side.

The lower pole of this kidney is moderately hydronephrotic and very free reflux occurs into it. The upper pole is healthy but transient reflux occurs into this also.

On the left side, he has a huge hydronephrotic lower half of the kidney remaining which did not reflux when the ureterocele was present but may do so now.

There was little doubt that this huge ureterocele was obstructing the outflow of the bladder and for this reason reflux preventing operations were not attempted.

On the 10.8.70, he was operated upon once more for left hyrocele and associated hernia. The scrotal sac was found to be full of fluid. There was a small soft hypoplastic testicle. The vas deferens was found to go down to the scrotum. But it did not connetc with the testicle.

X-ray examination did not reveal any vertebral abnormalities.

It is to be noted that most of the changes were to be found on the left side.

Another case of Goldenhaar syndrome was described in 1974 showed some vertebral abnormalities and a spine bifida.

Summary:

A case of Goldenhaar syndrome has been described.

In addition to the usual manifestations of the syndrome; there were extensive renal — and ureteric abnormalities and an inguinal hernia.

I would like to thank Mr. Williams of the Hospital for Sick Children for the report on the operations.

References

1 — Gross 1905: Ein Fall von agenesia Der Linken Lungen Beit: Path. Anat. 37-487 2 — Ocular manifestations of Goldenhaar's syndrome Oculo auriculo vertebral dysplasia.

Jules L. Baum and Murray Feingold.

XXI concilium ophthalmologium. Mexico 1970. Pars II pp 1361-1368.

3 — F.J. Damato. A case of oculo-auriculo-vertebral displasia

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