CASE REPORT

Case Report: Gollop-Wolfgang Complex in a 5 month old baby [v3; ref status: indexed, http://f1000r.es/53y]

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Abstract

Skeletal dysplasias are disorders associated with a generalized abnormality in the skeleton. The Gollop-Wolfgang complex (GWC) is a limb deficiency disorder and an unusual limb malformation with highly variable manifestations. Here we report an interesting case of a 5-month old male baby from India with Gollop-Wolfgang Complex showing bifurcation of the right femur, ectrodactyly of both feet, ectrodactyly of left hand, syndactyly of right hand and unusual presentation of bilateral fibular agenesis and caudal (Sacrococcygeal) agenesis. The etiology of GWC in this 5 month old male baby could possibly be attributed to spontaneous gene mutation. The clinical, radiographic findings and the unusual presentation are presented in detail.

This article is included in the Rare diseases channel.
Introduction
Generalized disorders of cartilage and bone have been referred to as skeletal dysplasias and are associated with a generalized abnormality in the skeleton. Gollop-Wolfgang Complex (GWC) is a rare congenital limb anomaly characterized by tibial aplasia, ipsilateral bifurcation of the thighbone and ectrodactyly. Ectrodactyly involves the deficiency or absence of one or more central digits of the hand or foot and is also known as split hand/split foot malformation (SHFM). Very often, the anomalies of limbs, heart, digestive and urinary tracts and the lumbosacral vertebrae are also affected.

In 1980, Gollop et al. described the case two brothers with ectrodactyly and unilateral bifurcation of the femur, absence of both tibiae and monodactyly of the feet. In 1984, Wolfgang reported a case of right femoral bifurcation and absence of tibia and bilateral central defects of the hand. Lurie and Ilyina (1986) proposed the eponym GWC for the combination of femoral bifurcation with hand ectrodactyly. Endo et al. found a total of 12 reported cases and added the case of a Japanese girl with a unique form of this malformation complex. Both hands and feet were involved and the involvement was bilateral. The etiology of GWC is most likely an error in the complex genetic control of limb development but the exact cause is still unclear. GWC is listed as a “rare disease” by the United States Office of Rare Diseases (ORD) of the National Institute of Health (NIH) and the approximate incidence is 1 in 1000,000.

Case presentation
A 5-month old male Indian child with normal karyotype (46 XY) born to a 26-year-old primigravida, full term by C-section, presented with limb deformities associated with bilateral ectrodactyly of feet (Figure 1 and Figure 2), syndactyly of right hand (Figure 3) and ectrodactyly of left hand (Figure 4). At the medial distal third of the right femur, a large protrusion was present (Figure 1 and Figure 5). Radiographic images showed bifid femur with fibular agenesis (Figure 6), absence of right 3, 4, 5 metatarsals and phalanges, absence of left 4, 5 metatarsals and phalanges of foot (Figure 7), left lateral X-ray showing caudal (sacrocccygeal) agenesis (Figure 8). Initial diagnosis was made when the parents brought the child to the out-patient department concerned about limb abnormality at the age of 3 months and the final diagnosis was made following admittance to the in-patient unit at 5 months, based on both clinical presentation and radiological images. There was no detailed prenatal history available.

The parents had documented second degree consanguinity but both did not have any significant family history. Similarly, there was no history of exposure to radiation, prenatal teratogenic medications and infections during pregnancy. The mother did not smoke or...
drink during pregnancy. The child was breast-fed with good appetite and cry, without any bowel bladder problems, change in skin color or any cleft lip/palate. Echocardiography at the time of admission revealed no congenital heart defects. The ultrasonography of abdomen and pelvis revealed no visceral or renal abnormalities. Surgical reconstruction treatment was advised but the parents did not give consent for treatment.

Discussion
One case was reported of an Arab Muslim couple who came from a region where other consanguineous families with similarly affected individuals had been reported Kohn et al. in 1989, and the autosomal recessive inheritance seemed evident in the case of a child described by Raas-Rothschild et al. in 1999. In this case, we report a typical presentation of GWC with bilateral fibular agenesis and sacrococcygeal agenesis along with pathognomonic features of GWC (bifurcation of femur, syndactyly and ectrodactyly). There were no associated abnormalities like cleft lip/palate, tibial agenesis,
visceral or cardiac anomalies seen in this patient. There is one documented case reported with distal femoral duplication with fibular agenesis. The best treatment option for this patient with GWC is early knee disarticulation and resection of the protruded bifurcated femur, followed by fitting of a modern prosthesis. This treatment was discussed with the parents of the patient at three months of age and a follow-up visit was scheduled after two months.

History of consanguinity is strongly associated with the developments of congenital anomalies among the newborn babies; there should be pre-marital genetic counselling to evaluate any impending congenital abnormalities. Similarly, antenatal check-ups are appropriate for early detection of congenital anomalies through proper screening.

Consent
Informed written consent for publication of images and clinical details was obtained from the patient’s parents.

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This report describes a 5-month-old patient with rare Gollop-Wolfgang Complex (GWC) disorder. The patient exhibits typical phenotypes of GWC disease, such as ectrodactylyl of the hand and bifurcation of the femur. The etiology of GWC remains unknown. However, the patient’s parents are second degree relatives, and other reports have described GWC patients in consanguineous families, suggesting that this disease is inherited in an autosomal recessive manner.

The manuscript is well-written, and I suggest it is indexed in current form.

We have read this submission. We believe that we have an appropriate level of expertise to confirm that it is of an acceptable scientific standard.

Competing Interests: No competing interests were disclosed.

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I have read this submission. I believe that I have an appropriate level of expertise to confirm that it is of an acceptable scientific standard.

Competing Interests: No competing interests were disclosed.
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I fully agree with the first reviewers's comments.

This paper deserves indexing as it is a very rare disorder. However, the following sentence (in the discussion) should be deleted:

"This type of skeletal dysplasia with limb deficiencies could be the result of spontaneous gene mutations and chronic exposure to a toxic substance or infectious agents that results in the disruption of normal skeletal development."

The main conclusion of this report should be that the occurrence of Gollop-Wolfgang in a consanguineous family brings further arguments to support autosomal recessive inheritance.

I have read this submission. I believe that I have an appropriate level of expertise to confirm that it is of an acceptable scientific standard.

Competing Interests: No competing interests were disclosed.
Author Response 02 Feb 2015

aatif qureshi,

Really appreciate your comments Dr. Stephen Robertson. I would like to hear back about my newer version of the paper.

**Competing Interests:** No competing interests were disclosed.