CASE REPORT

Two iniencephaly cases born the same day in the same hospital of Cali, Colombia. Iniencephaly cluster?

Wilmar Saldarriaga,¹ Carolina Isaza De Lourido,² Fabian Andres Ruiz Murcia³

¹Department of Morphology-Obstetrics and Gynecology, Universidad del Valle, Cali, Valle del Cauca, Colombia

²Department of Morphology, Universidad del Valle, Cali, Valle del Cauca, Colombia

³Department of Medicine and Surgery, Universidad del Valle, Cali, Valle del Cauca, Colombia

Correspondence to
Dr Wilmar Saldarriaga-Gil,
wilmar.saldarriaga@
correounivalle.edu.co

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SUMMARY

Two patients with short and hyperextended neck, cervical meningocele and other birth defects, were born the same day at the same hospital, both died. The autopsy report and X-rays show iniencephaly's classic findings. Revision of clinical histories did not show consanguinity between the parents, or exposure to environmental factors.

BACKGROUND

Iniencephaly is a rare neural tube defect of which no values exist of prevalence constructed with data from epidemiological surveillance with important numbers of births observed; values reported in the literature are established by isolated case series, extrapolated to the number of births in the population or maternities during short-time periods.

The birth of two cases with iniencephaly born on the same day at the same hospital is a very rare event, showing an unusual aggregation of cases and could suggest an iniencephaly's cluster.

This same hospital has reported high prevalence of some birth defects such as cyclopia and defects

due to vascular disruption, also a cluster of sirenomelia and cyclopia.

CASE PRESENTATION

Case 1

Mother, 20 years old (G2, A1); vaginal delivery at 35 weeks of gestation. Female newborn, weighing 1800 g, Apgar score of 2/10, deceased 10 min after birth. Autopsy reported 44 cm height, 30 cm cephalic perimeter, 26 cm thoracic perimeter, 23 cm abdominal perimeter; with broad nasal bridge, microstomia, hypoplasia of the mandible, low-set ears, hyperextended short neck with marked retroflexion of the head, with continuity of soft tissue, skin and hair between the back and head, abdominal excavation and hypoplasia of left lung. Dorsal-cephalic union; finding of cystic structure, concluding that it was a meningocele ([figure 1](#)); low-set ears, micrognathia ([figure 2](#)); X-ray showed

Figure 1 Case 1, lateral view. Note forced retroflexion of the head, producing a union of the occipital portion to the cervical and thoracic region of the spinal cord and meningocele.

Figure 2 Case 1, frontal view. Note hypoplasia of lower mandible and low-set ears.

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cervical hemivertebrae, marked retroflexion of the cervical cord and mandibular hypoplasia (figure 3).

Case 2

Primiparous mother 14 years old, caesarean delivery at 27 weeks of gestation. Newborn weighing 975 g, Apgar score of 1/10, deceased less than 5 min after birth. Postmortem findings: external genitalia of masculine characteristics, 25 cm height, 26 cm cephalic perimeter, 24 cm thoracic perimeter; hyperextended neck with forced retroflexion of the head, with union of the occipital portion to the cervical and thoracic region of the spinal cord; with continuity of soft tissue, skin and hair of the head to the back. Meningocele in dorsal-cephalic union (figure 4), low-set ears and micrognathia (figure 5). X-rays revealed: marked retroflexion of the cervical cord, fusion of vertebral-cervical bodies, thoracic hemivertebrae and hypoplasia of: clavicles, ulnas and radius (figure 6).

DIFFERENTIAL DIAGNOSIS

Iniencephaly exhibits similarity of the two entities: Kippel-Feil Syndrome (KFS), which is characterised by a short neck, low hairline and limited movement of the head, and is associated with deafness, congenital heart defects, mental deficiency and other abnormalities. KFS is caused by a failure in the segmentation of the cervical vertebrae in the fetal period, has been

Figure 3 Case 1, X-ray, lateral view. Note cervical hemivertebrae, marked retroflexion of the head over the whole spinal cord; in addition to mandibular hypoplasia.

Figure 4 Case 2, posterior view. Note forced retroflexion of the head, producing a union of the occipital portion to cervical and thoracic region of the spinal cord and meningocele.

classified in three types, type I with fusion of the cervical and upper thoracic vertebral bodies, type II affects two cervical bodies and type III with fusion of cervical and thoracic or lumbar vertebrae. KFS is rarely lethal and often can be corrected surgically.^{1 2}

The other similar phenotype is Spondylocostal dysostosis (SD) or Jarcho-Levin syndrome, characterised by short trunk with multiple vertebral and rib anomalies, can be associated with others defects, is generally not fatal.^{1 3} In both cases (KFS and SD), genetic counselling is needed, because there exists case reports with Mendelian inheritance patterns.¹⁻³

TREATMENT

Iniencephaly is generally associated with other anomalies such as cardiopathies, diaphragmatic hernia, intestinal atresia, renal defects, among others, making it a usually fatal condition¹; however, reports exist of patients with relatively mild conditions who have survived.⁴

DISCUSSION

Iniencephaly is a neural tube defect and is characterised by widening of the foramen magnum secondary to occipital bone defect, missing cervical or thoracic vertebrae, dysraphism, marked lordosis and hyperextension of the cervical cord, generating an upwardly oriented face and continuation of soft tissue between the back and the head, and jaw and thorax. The cause

Figure 5 Case 2, frontal view. Note hypoplasia of the lower mandible, low-set ears, deformed nose, short and winged neck.

Figure 6 Case 2, X-ray, lateral view. Note marked retroflexion of the head over the cervical cord, fusion of cervical-vertebral bodies, thoracic hemivertebrae and hypoplasia of: clavicles, ulnas and radius.

of iniencephaly is unknown and it is possible that it is heterogeneous.^{1 5}

The iniencephaly cases, reported, were found after the search and revision of cases with interesting congenital defects and/or congenital defects of rare occurrence born at the Hospital Universitario del Valle in Cali-Colombia, in photographic records belonging to gynaecology and obstetrics residents from Universidad del Valle. Case 1 was initially reported in a Colombian Journal.⁶ Afterwards the other case was found with similar characteristics and diagnosis of iniencephaly also with photographs, autopsy report, X-rays, medical history data of the mothers which were reviewed in detail. Birth records were checked, it was determined that both were born on the same day.

It has been described that cases of congenital defects of rare occurrence are observed during short periods of time, denominated as temporal aggregation. Different study methodologies are available to analyse these cases, seeking to establish if there is statistical significance between the number of cases observed and expected and if there is a common cause that can be intervened.^{7 8} This type of unusual appearance was reported over 2 decades ago for iniencephaly as a cluster in Miami with five cases in 4 months, since then there have been reports of an unusual appearance of such magnitude.⁹

The hospital where the cases occurred is located in the city of Cali, Colombia; which is the biggest public healthcare institution in the Colombian southwest, mostly caring for the lower income population in the region, attending approximately 6500

births per year. This institution belongs to the Latin American collaborative study on congenital malformations, ECLAMC—for the term in Spanish¹⁰ and since the implementation of epidemiological surveillance of congenital defects in 2004, it has reported a high prevalence of some congenital defects like cyclopia and defects due to vascular disruption^{11–13} including sirenómelia and cyclopia cluster.^{14 15} However, no case of iniencephaly was observed during the following 10 years.^{11–13}

The cases presented correspond to classical findings of iniencephaly; the two born at the same institution, the same day, which shows an unusual aggregation of cases and suggests the possibility of an iniencephaly cluster. The temporal and spatial analysis found approximately with a 2-month difference between the likely dates of conception; no coincidence was found between the places where the mothers spent their preconception period, case 1 in Cali, Department of Valle del Cauca, case 2 in Caloto, Department of Cauca; said municipalities are approximately 55 km apart from each other in a straight line, which is why the statistical analysis models for this aggregation would not be applicable; no evidence was found of kinship between the parents of the cases, finding no synonymy or exposure to environmental factors; only coinciding in being the product of young mothers (20 and 14 years of age) of low income, with this association being recognised for the appearance of greater congenital defects.¹⁶

The analysis determined that this aggregation of iniencephaly cases, in spite of being extremely rare, occurred randomly.

Learning points

- Iniencephaly is characterised by the presence of defect at the occipital osseous component, cervical or thoracic rachischisis and retroflexion of the head.
- Iniencephaly is a very low-prevalence neural tube defect.
- There was an aggregation of cases of iniencephaly without being a cluster.

Competing interests None.

Patient consent None.

Provenance and peer review Not commissioned; externally peer reviewed.

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