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Ritscher-Schinzel cardio-cranio-cerebellar (3C) syndrome: Report of three new cases. M. Leonardi, B. Wilkes, G. S. Pai. Department of Pediatrics, Medical Univ. of SC, Charleston.

Ritscher-Schinzel syndrome is also known as the 3C syndrome because it involves principally the cardiac, cerebellar and cranio-facial structures. However, a review of the 29 published cases shows considerable variability in its clinical manifestations. As many as half of all cases may involve only two of the three systems. Here we wish to report on three new cases, two of which were siblings diagnosed prenatally.

Cases 1 and 2. Baby R. was identified when autopsy studies following delivery at 31 weeks because of intrauterine fetal demise confirmed prenatally detected Dandy-Walker malformation, large atrial septal defect, cleft palate and hydronephrosis. During the subsequent pregnancy of this non-consanguineous, healthy Caucasian couple with a negative family history of malformations the fetus was found to have Dandy-Walker cyst, hypoplastic left heart, cleft palate and bilateral renal hypoplasia. Both fetuses had normal male karyotypes. Our third case is black male infant born at 38 weeks of gestational age to a 30-year-old primipara mother with no family history of birth defects or consanguinity. The infant weighed 1970 gm. at birth and was noted to have cleft palate, micrognathia, low set ears, tricuspid atresia with hypoplastic right ventricle, Dandy-Walker malformation and hydrocephalus. Other noted anomalies were a single umbilical artery, hypospadias, hemivertebrae and rib anomalies. Chromosomes were normal. The infant died following withdrawal of life support on the eighth day of life and no autopsy was performed. All three cases were seen during a one year period suggesting that the syndrome may be more common than is generally recognized.

We will present a detailed review of the 29 published cases and differential diagnosis of the 3C syndrome.

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VATER association with female hypospadia. J.L. Lin, C. J. Chen, J.Y. Lai and M. S. Hwang. Chang Gung Children's Hospital, Taoyuan, Taiwan.

A six months old female infant was found multiple anomalies and repeatedly admitted to our hospital after birth. Esophageal atresia with distal tracheo-esophageal fistula and duodenal atresia was diagnosed by X-ray and completely surgically repaired. Echocardiography showed truncus arteriosus type A1 with persistent ductus arteriosus. The hemodynamic status was temporally stabilized by B-T shunt creation. She had recurrent urinary tract infection. The renal echo showed severe right hydronephrosis and right ureter dilatation at birth which spontaneous regressed after subsequent follow-up. The VCUG showed that the urethral meatus located at anterior vaginal wall. We will present and discuss this rare urogenital condition- female hypospadia with VATER association.

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Fryns-Soekerman syndrome: Case report and review. D.R. McLeod[1,2] and C. Farr[2], [2]Department of Medical Genetics, [1]University of Calgary, and [2]Alberta Hereditary Diseases Program, Calgary, Alberta, Canada.

Fryns-Soekerman syndrome has been described in three previous males. X-linked inheritance appears likely based on only males being affected and minor stigmata present in two of the mothers. Karyotypes in all cases have been 46,XY with no evidence for micro-deletion on high resolution studies. In our case, the pregnancy was complicated by oligohydramnios and left ovarian dermoid cysts. At delivery, the infant was cyanosed and hypotonic and required ventilatory support. The birth weight was 2400 grams (<5th percentile) with a length of 44.5 cm (<5th percentile). He was noted to have severe micrognathia and a cleft palate in keeping with Pierre Robin sequence. At age 3 months, he was found to have a patent ductus arteriosus and on ECG had a Wolff-Parkinson-White type B conduction defect. At age 9 years, because of increasing neurologic problems, an MRI scan of the cranium and spine was done which showed small cerebellum, large cisterna magna and fourth ventricle and two areas of syringomyelia. Imaging studies of the spine showed the marked kyphoscoliosis and high vertebral bodies in relation to the AP diameter and only eleven ribs. Bone age was delayed. Radiographs of the hips showed bilateral coxa vara, shallow acetabula and superolateral subluxation of the femoral heads. On examination at 9 years, occipitofrontal circumference was 51.6 cm (40th percentile) with his weight below the 5th percentile. The frontal hairline was upswept and hair sparse. The ears had 15° of posterior rotation. The nasal bridge was prominent with a high insertion. The repaired cleft palate was narrow and elevated with a bifid uvula. There was moderate retrognathia with prominent large central incisors and hypodontia. Sweat pores on the dermal ridges were not reduced.

The mother had mild scoliosis which never required treatment but no other features suggesting she carried the gene. A maternal uncle had micrognathia and the maternal grandmother had scoliosis and a VSD. Review of family photographs did not suggest features of Fryns-Soekerman syndrome in these individuals.

We feel our case represents the fourth case of Fryns-Soekerman syndrome and expands the phenotype to include syringomyelia, eleven ribs and Wolff-Parkinson-White type B.

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Natural history of Trisomy 13 in unselected liveborn patients: Comparison with the S.O.F.T. data. E McPherson and M Clemens, Magee-Womens Hospital, Pittsburgh, PA and University of Pittsburgh.

Despite the frequency of trisomy 13, information regarding the natural history is limited. The frequently cited pessimistic survival statistics are based on the large (over 200 cases) study of Magenis et al.which is now 30 years old. More recently a survey of parents belonging to the Support Organization for Trisomies (SOFT) provided more optimistic survival data as well as some information on growth, medical history, and recurrence risk in trisomy 13 (Baty et al 1994). Because these data were provided by parents who belong to a self-selected group, the possibility of ascertainment bias toward milder, longer surviving cases must be considered.

We report our series of 18 unselected consecutive liveborn patients with trisomy 13 observed during the last 5 years. 10 patients died in the neonatal period, 1 died at 2.5 weeks, 1 died at 2.5 years, and the remainder were last known to be alive at ages of 1 mo, 3mo, 6mo, 1 yr, and 2 yrs (2 patients). Therefore the probability of survival was 47% at 1 week, 41% at one month, 33% at 6 mo, and 29% at 1 yr. Although the percentage of neonatal deaths is greater, the survival at later ages is comparable to the SOFT data. Complex congenital heart disease was the strongest indicator of poor prognosis, found in 7/9 patients who died neonatally and in none of those surviving more than 1 month. Other indicators of severe prognosis were gestation <36 weeks (5/9 neonatal deaths, 0 survivors) and severe CNS lesions (holoprosencephaly or neural tube defect) (4/9 neonatal deaths, 0 survivors). Of 9 infants who were ventilator dependent, only 1 was successfully weaned (after tracheostomy for tracheomalacia). Infants with trisomy 13 who are >36 weeks, not ventilator dependent, and do not have complex congenital heart disease or severe CNS lesions have a significantly better prognosis (5/6 survived >3months).

There were no familial recurrences of trisomy 13 in our study, but one family had a previous child with trisomy 18. The mothers of our patients had a total of 32 previous pregnancies resulting in 19 livebirths (including the trisomy 18) and 13 miscarriages (no reported theraputic abortions). These data are comparable to the SOFT data which included 40 previous pregnancies with 9 miscarriages and one family with a previous trisomy 18 conception. Combining these data yields a risk of 31% for miscarriage and 3% for autosomal trisomy in previous pregnancies. The recurrence risk for autosomal trisomy may be greater than previously suggested.