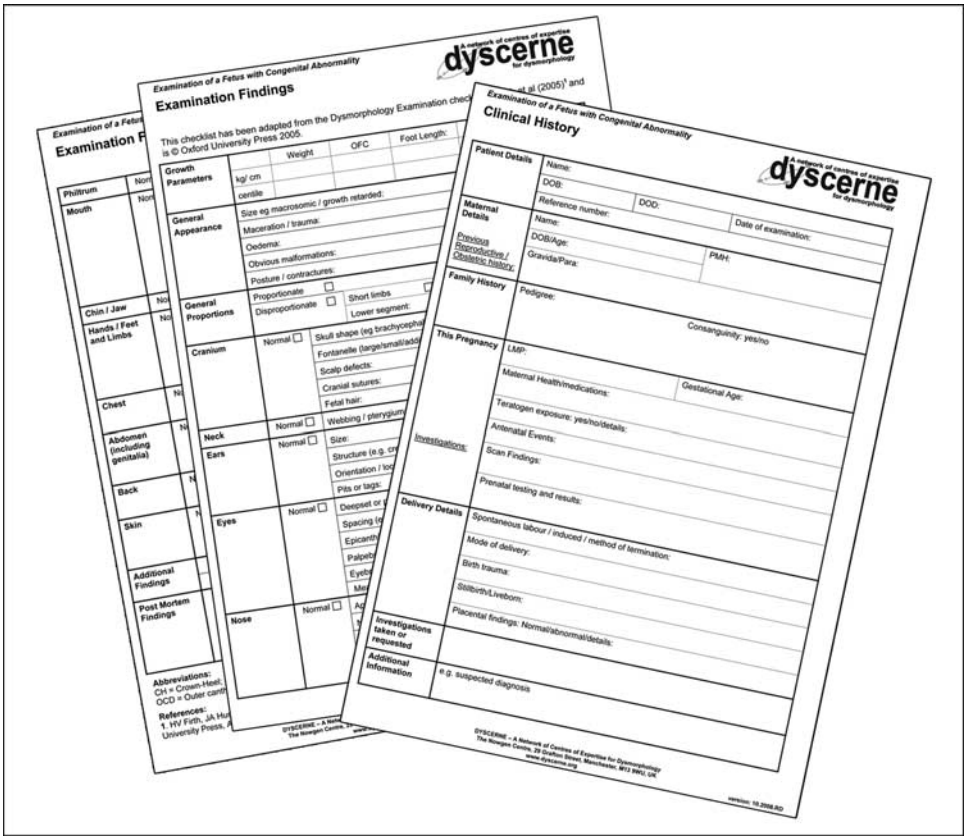
Fig. 2



Clinical history and examination findings proformas for the examination of a fetus with congenital abnormality.

and international networks for seeking expert advice, for example, DYSCERNE, and the European Skeletal Dysplasia Network (ESDN). Throughout this web tool, the emphasis is on consent, detailed observation, and documentation as well as interpretation of clinical findings in consultation with other experts. Its reference section is rather brief and could benefit from enabling direct links to the manuscripts listed. However, overall this is an excellent guide that will prove invaluable for all clinicians who perform external fetal examination but in particular those in training or involved on an infrequent basis.

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New syndrome – Situs inversus totalis with cystic dysplasia of kidneys, pancreas and bowing

Sevim Balcı[a], Fatih Ozaltin[b] and Sevinç Bostanoğlu[c], [a]Unit of Clinical Genetics, [b]Unit of Nephrology, Department of Pediatrics, Hacettepe University İhsan Doğramacı Children's Hospital and [c]Department of Radiology, Ankara Numune Hospital, Ankara, Turkey

Correspondence to Dr Sevim Balci, Professor Zirvekent 2.Etap C Blok No:58 K:8, D:35, Birlik Mah. Cankaya, Ankara, Turkey
Tel: +90 312 3051246; fax: +90 312 3094232;
e-mail: sbalci@hacettepe.edu.tr

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We read with interest a recent article in this journal 'Renal cystic dysplasia, paucity of bile ducts, situs inversus, bowing of the femora in two siblings in the Reunion Island: a ciliopathy' (Alessandri *et al.*, 2009). We would like to make a comment on this report. The authors discussed Ivemark syndrome, Cumming syndrome, and Jeune asphyxiating thoracic dysplasia to explain the features of their cases. However, we think that these cases are similar to the cases that we described as a new

autosomal recessive syndrome, which has been referred to as 'Situs inversus totalis with cystic dysplasia of kidneys and pancreas' in OMIM (#603643) (Balci *et al.*, 1999, 2000). We described three siblings with situs inversus totalis, cystic dysplasia of the kidneys, and pancreas that had been diagnosed prenatally in the 16th week of gestational age. We believe that the bowing of the femora described by Alessandri *et al.* (2009) may be a consequence of severe oligohydramnios and not a separate feature. In our patients, we performed full autopsy on all fetuses and detected no skeletal dysplasia as seen in Cumming syndrome or Jeune syndrome. Our syndrome differs from Ivemark syndrome because of the presence of situs inversus totalis and the absence of hepatic fibrosis and cysts. Hence the syndrome has been recognized as a new entity. Homozygosity was checked in the known loci for Meckel–Gruber syndrome (MEK1-3) and nephrocystin 2 in our patients. These loci were excluded as no evidence of homozygosity in the respective chromosomal regions was found. We looked for mutation by direct sequencing in the gene so-called 'intraflagellar transport 88 homolog isoform 2' (earlier known as Tg737), which has been suggested as a candidate gene for recessive renal diseases and as a modifier gene in human polycystic kidney disease (Onuchic *et al.*, 1995) as animal models exhibited lesions in the kidney, liver, and pancreas (Sommardahl *et al.*, 2001), and we found no mutation. After three affected siblings, the family had two healthy offspring (one male and one female), which supports autosomal recessive inheritance of the syndrome.

The cases described in the report of Alessandri *et al.* (2009) are very similar to our cases. Hence, we think that these cases may represent a variant of our syndrome rather than Cumming syndrome or renal–hepatic–pancreatic dysplasia.

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