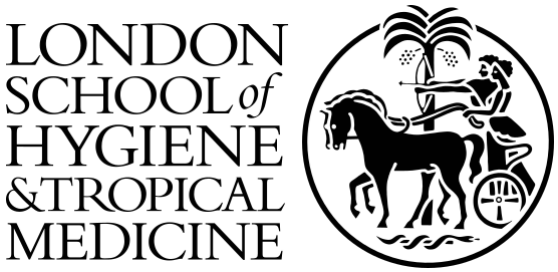

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TITLE PAGE

The most common comorbidities in Dandy-Walker Syndrome patients: a systematic review of case reports

Running title: Comorbidities in Dandy-Walker Syndrome

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Abstract

OBJECTIVE: Dandy-Walker Syndrome (DWS) is a rare neurological multi-entity malformation. This review aimed at reporting its main non-neurological comorbidities. METHODS: Following PRISMA guidelines, search in Medline was conducted (2000-2014, keyword dandy-walker). Age, sex, country, DWS type, consanguinity or siblings with DWS, and recorded coexistent conditions (by ICD10 category) were extracted for 187 patients (46.5% male, 43% from Asia) from 168 case reports. RESULTS: Diagnosis was most often set in <1y.o. (40.6%) or >12y.o. (27.8%). One-third of cases had a chromosomal abnormality or syndrome (N=8 PHACE), 27% had a cardiovascular condition (N=7 Patent Ductus Arteriosus), 24% had a disease of eye and ear (N=9 cataract); most common malignancy was nephroblastoma (N=8, all Asian). Almost one-fifth had a mental illness diagnosis; only 6.4% had mild or severe intellectual disability. CONCLUSION: The spread of comorbidities calls for early diagnosis and multidisciplinary research and practice, especially as many cases remain clinically asymptomatic for years.

Keywords: neuropathy, pediatric, neurodevelopment, developmental delay, cerebellum

MAIN TEXT

INTRODUCTION

Dandy-Walker Syndrome (DWS) is a rare congenital posterior fossa malformation, reported in only 1 in 25-30,000 live births, which occurs during the embryonic development of the cerebellum and 4th ventricle [1,2]. There is an ongoing debate among researchers when it comes to categorizing the variant forms of DWS; the most acceptable classification has been Barkovich's which identifies two types to the DW complex , A and B, wherein A split into the classic DW malformation and the DW variant, and B comprises the DW "mega cisterna magna" [1,3]. The aforementioned classification is now in dispute and some of these terms, like "variant" or "complex" , are largely avoided .Most cases of DWS are sporadic. However, it seems that first-degree relatives (such as siblings and especially twins) of DWS patients have increased risk of developing the condition compared to the general population, with the pattern of inheritance remaining unclear[4 5].

Diagnosis of the classic Dandy-Walker malformation is based on a series of characteristic neuroimaging findings, which include complete or partial agenesis of the cerebellar vermis, cystic dilatation of the 4th ventricle, enlarged posterior fossa. Hydrocephalus is a common complication of the disease (in almost 80% of the cases) [1,3]. Dandy-Walker malformation could be diagnosed before birth sonographically (by identifying certain sonographic features such as the size of ventricles or measurements of the brainstem- vermis angle) [6,7] but the majority of cases seem to be diagnosed postnatally and, in fact, before the age of 1-year-old.

DWS has multiple neurodevelopmental complications since the cerebellum, which is the mainly affected structure, is the region of the brain that regulates movement coordination as well as, partially, cognition and behavior [8].The symptoms usually become apparent in early infancy with the most common being macrocephaly, signs of increased intracranial pressure and spastic paraparesis , hypotonia, slow motor development and intellectual disability can also be observed in the early stages . Other less common signs include focal neurologic signs such as strabismus, nystagmus and palsies of cranial nerves , truncal ataxia and speech difficulties [1,9,10]. Some patients with DWS may also suffer from seizures which are usually associated with supratentorial malformations [11].

DWS comorbid with non-neurological clinical entities can be a treatment challenge and is currently under research .Many cases of DWS have been associated with major psychopathology such as schizophrenia [12], bipolar disorder [13], major depression and impulse control disorder [14]. Further, a seemingly high percentage of children with DWS have dysmorphic characteristics [15], other malformations of the Central Nervous System (agenesis of the corpus callosum, rachischisis, ectopic brain or cerebellar tissue) [1,16], as well as disorders of the cardiovascular [17], urogenital [18], and gastrointestinal systems [19]. Multiple types of neoplasms and neurocutaneous disorders have also been observed in variable frequencies [20,21]. In addition, other genetic syndromes [22] and chromosomal anomalies, such as 3q Syndrome (the location of the DWS affected genes, known as ZIC1 and ZIC4) [1,23] and trisomy 18 [24] may coexist. However, despite many observations, the sporadic nature of the disease and the expectation that it is now relatively well understood in Europe and North America has impeded research in large case-series to explore clinically significant

comorbidities and most of the above observations are derived from low evidence case reports.

The aim of this paper has been to systematically review the literature, collect all disparate case reports of children with all types of DWS into a secondary case series and report on the frequency of non-neurological medical conditions coexisting with DWS.

METHODS

Working definitions

For the purposes of this review, comorbidity was defined as a systemic medical condition existing simultaneously with DWS in a single patient, regardless of the existence of a reported causal relationship [25]. Any neuropathological and morphologic abnormalities were excluded. For the classification of comorbidities, the 10th revision of the International Statistical Classification of Diseases and Related Health Problems (ICD10 list) was used. As outlined above, DWS has been considered as an all-entity encompassing term.

Search strategy

This review has adopted the Preferred Reporting Items for Systematic Reviews and Meta-Analyses (PRISMA) guidelines [26]. Using only variations of the main keyword "dandy-walker", a systematic search was done in PubMed from 01/2000 to 05/2014 by two reviewers working independently. Inclusion criteria comprised the following: English-language publications, case reports or small case series and publications reporting comorbidities or coexisting conditions associated with Dandy-Walker. All other study designs, secondary research, and studies reporting on diagnosed fetuses who did not survive to birth were excluded. Two reviewers independently screened titles and abstracts of retrieved citations. The full texts of potentially relevant case reports were screened. A standard search diary was kept by each reviewer. Any disagreements were resolved through consensus following consultation with a third reviewer. A relevant flow chart was constructed to outline the number of papers retrieved and exclusions in each step.

Data extraction and synthesis

Three reviewers extracted the following information from each study on an Excel® speadsheet: study title, first author's surname, year of publication, study design, country of origin of patient, sex, age group (according to the Centers for Disease Control and Prevention Child Development classification), Dandy-Walker type (syndrome/malformation, variant and complex/mega cisterna magna, as defined in each article), parents or siblings diagnosed with DWS. To ensure that the review was inclusive of all relevant findings, studies

which reported patients with Dandy-Walker anomaly were included even when they were not confirming the diagnostic radiological evidence. In the event of missing data, additional information was requested via contacting the corresponding authors.

Most variables were expressed qualitatively. A purely descriptive approach was adopted (i.e. data expressed as non-weighted means whenever possible) for continuous variables. No further statistical analysis was undertaken. In the interest of not excluding clinically important case reports, no formal quality assessment of eligible articles was undertaken.

RESULTS

Literature search

The search yielded a total of 576 papers (Figure 1). Two-hundred forty-nine papers were excluded by screening of the title and abstract; 195 were irrelevant, 48 were non-English publications and 6 reported on fetuses or stillborn children. Following screening of the full text for 326 publications, 168 case reports were eventually included , which represented primary case reports or reports of cases in two siblings, corresponding to a total of 187 patients.

Study characteristics

Characteristics of the included case reports are summarized in Table 1. In most included cases the diagnosis of Dandy-Walker Syndrome was established by Magnetic Resonance Imaging (MRI) of the brain. Eighty seven (46.5%) individuals were male and 80 (42.8%) were female, while 20 (10.7%) studies did not report the gender of the case. At the point of diagnosis, 52 patients (27.8%) were 12 years old or older, 14 patients (7.5%) were between 5-12 years old, 29 (15.5%) were toddlers or of preschool age (1 -5 years old) and 76 patients (40.6%) were infants or neonates (younger than 1 year old); age was not reported in 16 cases (8.6%).

As far as area of origin is concerned, most of the patients (43%, N=81) came from Asia, approximately a quarter (28%, N=52) from Europe, a further 16% (N=29) from North America, and the rest from South America (N=14), Africa (N=8), and Oceania (N=2). Most cases reported from a single country were from the United States (N=24), followed by Turkey (N=20), India (N=18), Japan (N=16), with Germany contributing a fifth of European cases (N=11).

Fifteen patients had a parent diagnosed with DWS (16%), whereas in half of the studies (49.7%, N=93 patients) this was not reported . Twenty cases had one or more siblings with Dandy- Walker malformations (21%). There was no evidence regarding siblings with DWS in 95 cases.

Most of the patients (64.7%, N=121) had been diagnosed with Dandy Walker malformation (or syndrome), 37 patients (19.8%) were reported as having the less severe form of Dandy

Walker variant, 23 (12.3%) patients had Dandy-Walker complex, 2 (1.1%) had mega cistern magna. Dandy Walker anomaly was recorded as part of the Dandy Walker Syndrome. The terms “variant” and “complex” were recorded for 62 cases to indicate disease type and severity, despite them being terms not widely used currently in Europe and North America.

Comorbidities

Comorbidities reported in all case reports combined, classified by ICD10 category, are summarized in Table 2. The most frequent ones are outlined below.

Neoplasms: One or more malignancies were reported in 22 patients (11.8%), most Asian (N=144, 77%) . Nephroblastoma (Wilm’s tumor) was the most frequent, diagnosed in 8 patients (4.4%) from Asia. Four patients (2.1%) had tongue hamartoma.

Endocrine and metabolic diseases: Overall, 18 patients (9.6%) were reported to have diagnosed endocrine/metabolic disorder, with hirsutism being the most common(N=5, 2.7%).

Mental health and learning disability: Thirty-one patients (16.6%) were diagnosed with a mental or behavioral disorder, almost half of them being European (N=14, 45%) and a third (N=11, 35%) Asian, with an approximately equal number of women and men. About half (N=16, 51%) of these patients had Dandy Walker variant while a third (N=9, 29%) had Dandy Walker malformation and fewer (N=6, 19%) Dandy Walker complex. Twelve of them (38.7%) also had a diagnosed learning disability (severe or mild), 5 (16.1%) had bipolar disorder, 5 (16.1%) had symptoms of psychotic spectrum, while 4 patients (12.9%) were reported to have Attention Deficit/Hyperactivity Disorder (ADHD).

Diseases of the eye and ear: Twenty-four per-cent (N=45) of the patients had one or more ocular abnormalities and almost half of them (N=20, 44%) were Asian and a third (N=14, 31%) were European. Five (11%) had a diagnosed parent, while 10 (22%) had one or more siblings with DWS. Nine patients (20%) had a diagnosed cataract (almost half, N=4, 44.4%, of them had at least one sibling with Dandy Walker), 6 (13.3%) had microphthalmia, 5 patients (11.1%) had chorioretinal atrophy /dysplasia, 5 (11.1%) had optic nerve dysplasia/atrophy, 5 (11.1%) had microcornea/corneal opacity and 5 (11.1%) had reported myopia. Three patients (6.7%) were diagnosed with coloboma of the eye (iris/optic nerve/optic disk) and 7 patients (15.6%) had hearing impairment.

Diseases of the circulatory system: Overall, 27% (N=51) of the patients had a diagnosed heart or vascular disorder. Most of them were females (N=32, 62%), almost half of them were Asian (N=22, 43%) and a quarter (N=13, 25%) were European. The most common heart problem was patent ductus arteriosus (PDA) diagnosed in 7 patients (13.7%), 6 patients (11.8%) had coarctation of the aorta, 6 (11.8%) had ventricular septal defect and 5 (9.8%) had atrial septal defect (ASD). Heart failure was reported in five (9.8%) patients.

Diseases of the skin and subcutaneous tissue: Skin conditions were reported in 37 patients (19.8%). Neurocutaneous melanosis was the most common - observed in 11 (29.7%) of these patients - followed by hemangiomas (9 patients, 24.3% – including those who suffered from PHACE syndrome) and melanocytic nevi (N=6, 16.2%).

Diseases of the musculoskeletal system and connective tissue: Sixteen patients (9%) were reported to have musculoskeletal comorbidities; this was almost exclusively observed in patients with Dandy Walker malformation. Four of them (25%) had scoliosis/kyphoscoliosis while joint laxity was reported in three patients (18.8%).

Diseases of the genitourinary system: A total of 26 patients (14%) included in this review had mild or severe genitourinary problems. Ten patients (38.5%) had kidney malformations (polycystic kidneys/dysplastic kidney/agenesia/pelvic kidney), 7 (26.9%) had hypoplastic genitalia (poorly formed uterus/micropenis/concealed penis), and 4 (15.4%) had cryptorchidism/undescended testis.

Syndromes and chromosomal abnormalities: Approximately one third of the patients (N=62) were described to have a syndrome or chromosomal abnormality. A tenth of these patients had a parent diagnosed with DWS. The most common syndrome was PHACE Syndrome, which was reported in eight cases. Four patients (6.5%) had Edwards Syndrome (trisomy 18) and 4 (6.5%) had Joubert Syndrome.

DISCUSSION

Summary of findings

Dandy-Walker syndrome is a congenital malformation which is associated with a complex set of neurological as well as non-neurological medical conditions, as this review clearly demonstrated.

The review built up a case series of 187 patients, potentially allowing for some generalizations as well. The reported diagnosed patients were most often male, which contradicts older studies [27]. Interestingly, almost half of the patients were diagnosed in the first year of life, which is consistent with published literature [28], however, the second peak for diagnosis (over a quarter of cases) was in young people older than 12 years old, where the syndrome is suspected usually due to mental health problems. As far as types are concerned, malformation was the most frequent, followed by variant, with the rest only being reported very rarely. A clear increased prevalence among Asian populations was extrapolated from this review; this may reflect poor access to prenatal diagnostic tests in certain settings and/or decrease in publication of cases from Europe (of note, most cases from a single country where from the US). Contrary to previous studies [29] [30], no clear statistically important correlation between DWS in the offspring and clinical disease of the parents was found.

Comorbidities

The review resulted in interesting findings on non-neurological comorbidities. Of note, approximately a third of the patients with DWS, were reported to have a chromosomal abnormality or another genetic syndrome, the most frequent being PHACE Syndrome,

trisomy 18 and Joubert Syndrome, as has been described in the literature [31,32]. Several malformation syndromes and cytogenetic abnormalities have been associated with Dandy Walker malformation and several genes have recently been implicated in its pathogenesis. These include ZIC1 and ZIC4 of chromosome 3q24 and it seems that the deletion of these genes in mice could account for associated defects similar to Dandy Walker malformation and differences in cerebellar size [32-34]. FOXC1, FGF17, LAMC1 and NID1 have also been associated with DWS[2]. Several published reports have suggested that, there is a causal relationship between various types of chromosomal abnormalities and malformation syndromes [32]. In this review, it was indeed identified that trisomy 18 was the most frequent chromosomal abnormality and this is consistent with the literature as DWS has been reported to be a frequent complication of trisomy 18 and therefore, imaging tests should be conducted at an early stage [32]. It is more difficult to explain the potential association with PHACE Syndrome (Posterior fossa malformations–Hemangiomas–Arterial anomalies–Cardiac defects–Eye abnormalities–sternal cleft and supraumbilical raphe syndrome) which is a cutaneous condition linked to multiple congenital abnormalities. Although its pathogenesis, which implicates the neural crest, includes cerebellar involvement, the full mechanism has not been fully clarified yet [35].

The second most common group of comorbidities, occurring in a quarter of patients, was cardiovascular conditions, which is also consistent with the literature [17]. The basis for this association could well be in the suggested timing of the formation of DW malformation with a complex embryologic defect originating before the 6th week of gestational development. Given the significant number of patients with a heart defect, some of which may lead to heart failure, monitoring and awareness are important for the prognosis, especially in case of a shunting procedure[1]. A recent published case report also highlighted the important association between DWS and vascular disease [36].

Other conditions reported in high numbers included diseases of the eye and ear and skin conditions, the most common being neurocutaneous melanosis. This is considered to follow from disorders in neurulation, the same that may cause the development of malformations like DWS. Meningeal cells are known to play a critical role in the neuromorphogenesis of the cerebellum and abnormal leptomeninges infiltrated by melanocytic cells may interfere with normal cerebellar development[37]. The exact pathogenesis of DWS in this association, however, cannot be fully explained yet. In a different non-prevalent theory, melanocytic cells may cause atresia or obstruction in the outgoing foramen of the fourth ventricle leading to cystic dilatation [38].

The prevalence of mental health problems was an important finding of this review. Notably, most patients with mental/behavioral problems were from Europe, were diagnosed with variant and were older than 12 years, evidence that is not dissimilar to previous publications[39]. This is not surprising as cognitive, affective and behavioural changes in psychiatric disorders have been linked to structural cerebellar abnormalities [40], including schizophrenia in which the cerebellum's role in cognition, particularly the role of the vermis, is very important [41]. Further, it is well known that patients diagnosed with ADHD disorder are unable to delay or inhibit a response, which is considered to be a central executive function, and cerebellum is one of the structures modulating this. Several studies have

already pointed toward cerebellar deficits in ADHD, and vermal volume was consistently found to be significantly smaller in children with this disorder[35,39]. The key role of mental health problems in the prognosis of DWS patients is also clarified through three recent case reports which are mainly focused on bipolar disorder and mild cognitive impairment[42-44].

Lastly, results on intellectual development are inconsistent. Some studies have reported that the majority of these children have a learning disability[45], with others suggesting that most of them have IQ in the normal range [6,27]. Of note only 6.4% of DWS patients had mild or severe intellectual disability in our review

. Any extra-CNS diseases that coexist with DWS should be detected early, given that they play an important role in the development and health outcomes of these children [45,46] . Postnatal outcomes vary from normal psychomotor development to severe disability or death, which highlights the importance of early diagnosis[47].

Strengths and Limitations

This review is presented as a proxy for reporting results on one of the largest cohorts of DWS patients ever pulled together in one study. It benefits from a strong standard methodology, and exhaustive reporting of all comorbidities in every of 187 patients reported. However, limitations to this review should be acknowledged. For reasons of access, only one database (Medline) was searched in one language (English). Second, only information referring to alive patients with Dandy-Walker Syndrome was included. Third, it should be noted that, given that there are no concrete classification criteria for the different types of DWS, outcomes reported may vary in different settings and studies, thus resulting in heterogeneity of reporting. This is further complicated by the rarity of the condition which suggests that most clinicians-authors would not be very familiar with its manifestations. Moreover, the fact that almost half of the case reports originate from Asian countries potentially leads to some degree of selection bias, though we were unable to extract from the literature if there has been a decrease in the interest of publishing such case reports in well-resourced settings like in North America. . Further, as mentioned previously and in an attempt to be inclusive, we included some cases which were recorded as “DW variant” or “complex” (terms not in wide use any more) as these terms have been used consistently by clinicians and researchers alike for several years. Finally, it should be noted that the comorbidities which were part of the syndromes described in this study were not reported separately and therefore, we could assume that certain medical conditions coexist more frequently with DWS.

Conclusion

Results from a secondary cohort of 187 patients with Dandy-Walker Syndrome highlight various genetic, cardiac and mental health abnormalities of importance as comorbidities to the syndrome, while clarifies the – previously thought higher – rates for intellectual disability

and cancer. Given the rarity of the condition internationally, future research should more realistically focus on exploring the pathophysiological mechanisms between some of these correlations to lead into targeted treatment and prevention. Comorbidities impact heavily on the prognosis and quality of life of patients, thus early diagnosis (over a quarter of DWS cases are still diagnosed after the age of 12 years) and multi-specialist approaches based on these epidemiological findings could help towards making a difference for this patient group, especially as many DWS cases lead a subclinical course for many years.

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Author Contributions:

ES: literature search, data extraction, data analysis, text drafting, text editing, final manuscript approval

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KK: data interpretation, critical revision of manuscript, text editing, final manuscript approval

MD: data extraction, data interpretation, text drafting, text editing, final manuscript approval

AAK: study concept, study design, critical revision of manuscript, text editing, final manuscript approval

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Figure 1.Flow chart

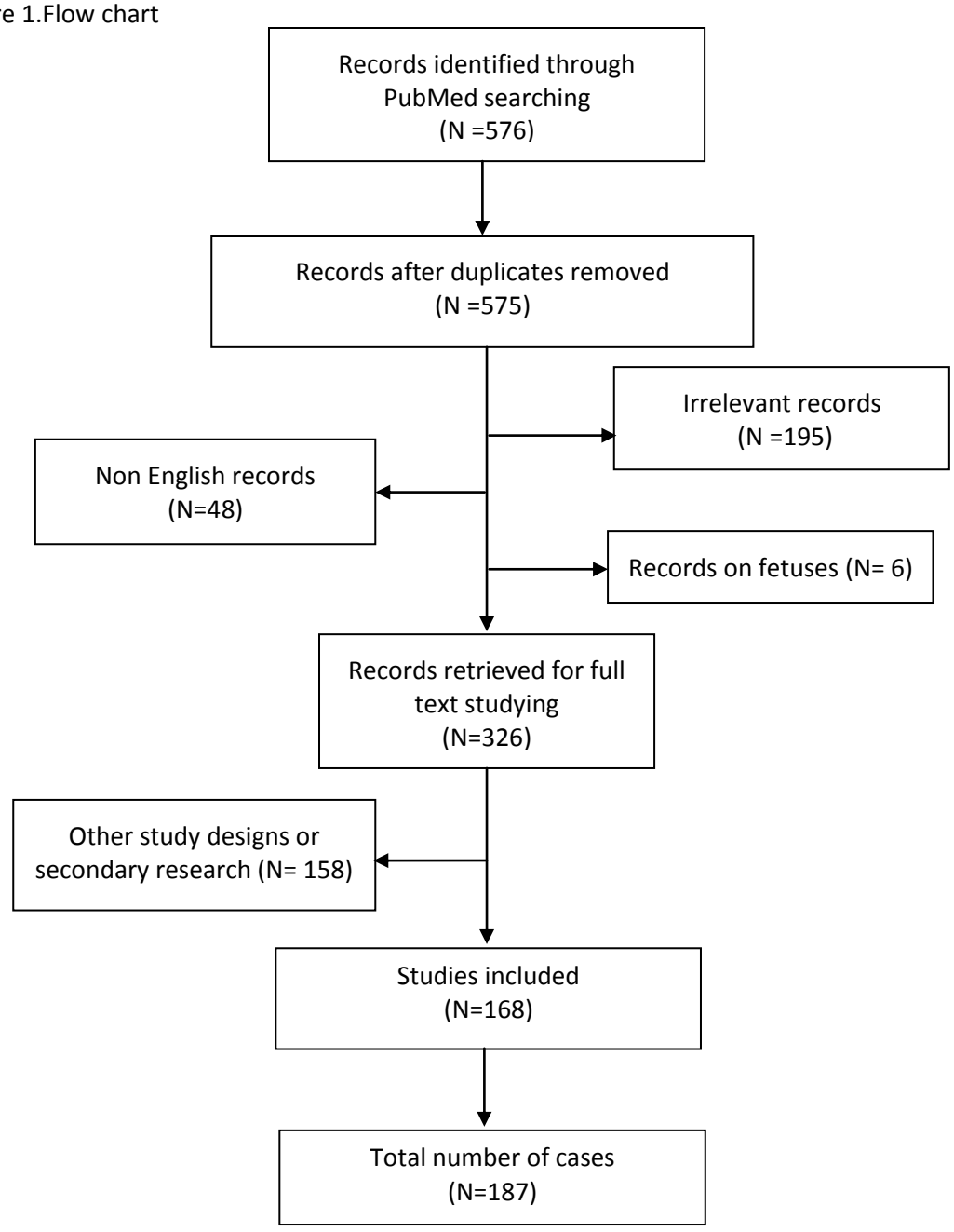

Table 1: Baseline characteristics of all included studies

Study Name	Continent	Sex	Age group	Type	Consanguinity	Siblings with DW
Walch et al, 2000[1]	Europe	F	1-12m	Malformation	No	No
Koul et al, 2000[2]	Asia	M	13m-3y	Syndrome	*N/A*	Yes
Koul et al, 2000[2]	Asia	M	13m-3y	Syndrome	*N/A*	Yes
Pallotta et al, 2000[3]	Europe	F	5-12y	Variant	*N/A*	*N/A*
Matsuura et al, 2000[4]	Asia	M	1-12m	Malformation	*N/A*	*N/A*
Matsuura et al, 2000[4]	Asia	M	13m-3y	Malformation	*N/A*	*N/A*
Poetke et al, 2000[5]	Europe	*N/A*	13m-3y	Malformation	*N/A*	*N/A*
Poetke et al, 2000[5]	Europe	*N/A*	13m-3y	Malformation	*N/A*	*N/A*
Poetke et al, 2000[5]	Europe	*N/A*	13m-3y	Malformation	*N/A*	*N/A*
Haug et al, 2000[6]	Asia	F	1-12m	Malformation	No	Yes
Haug et al, 2000[6]	Asia	M	1-12m	Malformation	No	Yes
Ben Hamouda et al, 2001[7]	Africa	M	1-12m	Syndrome	*N/A*	*N/A*
Arslanoglu et al, 2001[8]	Asia	*N/A*	*N/A*	Complex	Yes	Yes
Gulcan et al, 2001[9]	Asia	M	*N/A*	Malformation	*N/A*	*N/A*
Fan et al, 2001[10]	North America	M	<1m	Malformation	No	No
Lapunzina et al, 2001[11]	South America	F	<1m	Malformation	No	No
Gilbert- Barness et al, 2001[12]	North America	M	<1m	Malformation	No	No
Sudha et al, 2001[13]	North America	F	5-12y	Variant	No	No
McKee et al, 2001[14]	Asia	F	3-5y	Malformation	Yes	No
Imai et al, 2001[15]	Asia	M	1-12m	Variant	No	No
Kajii et al, 2001[16]	Asia	M	1-12m	Complex	No	No
Kajii et al, 2001[16]	Asia	F	3-5y	Complex	No	No
Kajii et al, 2001[16]	Asia	F	1-12m	Complex	No	No
Kajii et al, 2001[16]	Asia	F	13m-3y	Complex	No	No
Kajii et al, 2001[16]	Asia	M	13m-3y	Complex	No	Yes
Kajii et al, 2001[16]	Asia	M	1-12m	Complex	No	Yes
Gulati et al, 2002[17]	Asia	M	1-12m	Complex	No	No
Kisato et al, 2002[18]	Asia	M	1-12m	Malformation	*N/A*	*N/A*
Yoder et al, 2002[19]	North America	M	13m-3y	Malformation	*N/A*	*N/A*
Wieselthaler et al, 2002[20]	Africa	M	5-12y	Variant	No	No
Mena-Cedillos et al, 2002[21]	North America	M	3-5y	Malformation	No	No
Hadzikaric et al, 2002[22]	Asia	M	13m-3y	Syndrome	*N/A*	*N/A*

Hammond et al, 2002[23]	Europe	M	>12y	Complex	*N/A*	*N/A*
Peters et al, 2002[24]	Europe	M	13m-3y	Malformation	*N/A*	*N/A*
Freeman et al, 2002[25]	Europe	M	>12y	Syndrome	*N/A*	*N/A*
Toriello et al, 2002[26]	North America	F	1-12m	Malformation	*N/A*	*N/A*
Aslan et al, 2002[27]	Asia	M	<1m	Malformation	Yes	No
Lin et al, 2003[28]	Asia	F	1-12m	Malformation	*N/A*	*N/A*
Furukawa et al, 2003[29]	Asia	*N/A*	*N/A*	Malformation	*N/A*	*N/A*
Dinakar et al, 2003[30]	Europe	F	5-12y	Malformation	Yes	No
Panteliadis et al, 2003[31]	Europe	F	13m-3y	Malformation	*N/A*	*N/A*
Hedera et al, 2003[32]	North America	M	<1m	Malformation	*N/A*	*N/A*
Litzman et al, 2003[33]	Europe	F	>12y	Syndrome	No	No
Erdal et al, 2003[34]	Asia	*N/A*	*N/A*	Complex	*N/A*	*N/A*
Cazorla Calleja et al, 2003[35]	Europe	F	1-12m	Malformation	No	No
McCormack Jr et al, 2003[36]	North America	*N/A*	*N/A*	Malformation	*N/A*	*N/A*
McCormack Jr et al, 2003[36]	North America	*N/A*	*N/A*	Malformation	*N/A*	*N/A*
Muzumdar et al, 2004[37]	Asia	F	1-12m	Malformation	No	No
Arai et al, 2004[38]	Asia	*N/A*	*N/A*	Malformation	*N/A*	*N/A*
Mohammadi et al, 2004[39]	Asia	F	1-12m	Malformation	*N/A*	*N/A*
Tubbs et al, 2004[40]	North America	F	13m-3y	Variant	*N/A*	*N/A*
Ueno et al, 2004[41]	Asia	M	>12y	Malformation	*N/A*	*N/A*
Owler et al, 2004[42]	Oceania	F	>12y	Malformation	*N/A*	*N/A*
Prieto Espuñes et al, 2004[43]	Europe	F	<1m	Malformation	*N/A*	*N/A*
Weinzieri et al, 2005[44]	Europe	*N/A*	<1m	Malformation	*N/A*	*N/A*
Ross et al, 2005[45]	Oceania	F	1-12m	Malformation	No	No
Castro Conde et al, 2005[46]	Europe	M	<1m	Variant	No	No
Sun et al, 2005[47]	North America	F	<1m	Malformation	No	No
de Azevedo Moreira et al, 2005[48]	South America	F	1-12m	Malformation	No	No
Teksam et al, 2005[49]	Asia	F	1-12m	Malformation	Yes	No

Ounap et al, 2005[50]	Europe	F	<1m	Malformation	No	No
Iyer et al, 2005[51]	N/A	F	*N/A*	Malformation	*N/A*	*N/A*
Klein et al, 2005[52]	North America	M	5-12y	Malformation	No	No
Pascual-Castroviejo et al, 2005[53]	Europe	*N/A*	*N/A*	Complex	No	No
Pascual-Castroviejo et al, 2005[53]	Europe	*N/A*	*N/A*	Complex	No	No
Kolomietz et al, 2005[54]	North America	F	1-12m	Malformation	No	No
Prasad et al, 2006[55]	Asia	*N/A*	<1m	Malformation	*N/A*	*N/A*
Weimer et al, 2006[56]	Europe	F	<1m	Variant	*N/A*	No
Menon et al, 2006[57]	Asia	F	>12y	Malformation	*N/A*	*N/A*
Kim et al, 2006[58]	Asia	M	>12y	Malformation	No	No
Chen et al, 2006[59]	Asia	M	5-12y	Variant	*N/A*	*N/A*
Cappellacci et al, 2006[60]	Europe	M	5-12y	Malformation	No	No
Kang et al, 2006[61]	Asia	M	*N/A*	Malformation	*N/A*	*N/A*
Richter et al, 2006[62]	North America	F	<1m	Malformation	*N/A*	*N/A*
Kalayci et al, 2006[63]	Asia	F	<1m	Malformation	No	No
Akgul et al, 2006[64]	Asia	F	>12y	Variant	*N/A*	*N/A*
Cannady et al, 2006[65]	North America	F	<1m	Malformation	*N/A*	*N/A*
Caceres et al, 2006[66]	North America	F	1-12m	Complex	*N/A*	*N/A*
Abdel-Salam et al, 2006[67]	Africa	M	3-5y	Malformation	Yes	Yes
Abdel-Salam et al, 2006[67]	Africa	M	13m-3y	Malformation	Yes	Yes
McPherson et al, 2006[68]	North America	M	<1m	Malformation	No	No
Su et al, 2007[69]	Asia	F	<1m	Malformation	*N/A*	*N/A*
McClelland et al, 2007[70]	North America	M	1-12m	Complex	*N/A*	*N/A*
De Brito Henriques et al, 2007[71]	South America	F	*N/A*	Syndrome	*N/A*	*N/A*
Sener, 2007[72]	Asia	F	1-12m	Malformation	Yes	*N/A*
Aluclu et al, 2007[73]	Asia	M	>12y	Malformation	*N/A*	*N/A*
Papazisis et al, 2007[74]	Europe	M	>12y	Variant	*N/A*	*N/A*
Poot et al, 2007[75]	Europe	M	5-12y	Complex	No	No
Cardoso et al, 2007[76]	South America	F	>12y	Syndrome	No	No
Ghosh et al, 2007[77]	Asia	F	<1m	Malformation	No	No
Humphries et al, 2007[78]	North America	F	>12y	Variant	*N/A*	*N/A*
Vundinti et al,	Asia	M	3-5y	Malformation	No	*N/A*

2007[79]						
Ferentinos et al, 2007[80]	Europe	F	>12y	Mega Cisterna Magna	*N/A*	*N/A*
Zechi-Ceide et al, 2007[81]	South America	*N/A*	*N/A*	Variant	No	Yes
Zechi-Ceide et al, 2007[81]	South America	*N/A*	*N/A*	Variant	No	Yes
Zechi-Ceide et al, 2007[81]	South America	*N/A*	*N/A*	Variant	No	Yes
Hou et al, 2008[82]	Asia	M	1-12m	Variant	No	No
van Steensel et al, 2008[83]	Europe	M	>12y	Malformation	No	No
Herman et al, 2008[84]	North America	M	1-12m	Malformation	*N/A*	*N/A*
Saatci et al, 2008[85]	Asia	M	13m-3y	Variant	No	No
Cakmak et al, 2008[86]	Asia	M	<1m	Malformation	*N/A*	*N/A*
Schreml et al, 2008[87]	Europe	F	5-12y	Malformation	*N/A*	*N/A*
Warwick et al, 2008[88]	North America	F	>12y	Malformation	*N/A*	*N/A*
Cushing et al, 2008[89]	North America	F	1-12m	Malformation	*N/A*	*N/A*
Kasliwal et al, 2008[90]	Asia	*N/A*	13m-3y	Malformation	*N/A*	*N/A*
Kurdi et al, 2009[91]	Asia	M	1-12m	Malformation	Yes	*N/A*
Nagdeve et al, 2009[92]	Asia	F	1-12m	Malformation	*N/A*	*N/A*
Caglayan et al, 2009[93]	Asia	F	1-12m	Malformation	No	No
Venturini et al, 2009[94]	Europe	M	>12y	Malformation	*N/A*	*N/A*
Murugesan et al, 2009[95]	Asia	M	>12y	Malformation	*N/A*	*N/A*
Marnet et al, 2009[96]	Europe	M	>12y	Complex	*N/A*	*N/A*
Lingeswaran et al, 2009[97]	Asia	M	5-12y	Variant	*N/A*	*N/A*
Cultrera et al, 2009[98]	Europe	M	>12y	Variant	*N/A*	*N/A*
Linder et al, 2009[99]	North America	M	1-12m	Complex	No	No
Gönül et al, 2009[100]	Asia	F	>12y	Malformation	No	No
Ozdemir et al, 2009[101]	Asia	F	1-12m	Variant	Yes	No
Walbert et al, 2009[102]	North America	F	>12y	Malformation	*N/A*	*N/A*
Masdeu et al, 2009[103]	Europe	M	>12y	Malformation	*N/A*	*N/A*
Zhang et al, 2009[104]	Asia	F	>12y	Malformation	No	No

Sato et al, 2009[105]	North America	M	>12y	Malformation	*N/A*	*N/A*
Surányi et al, 2009[106]	Europe	M	1-12m	Malformation	*N/A*	*N/A*
Al-Adnani et al, 2009[107]	Europe	*N/A*	*N/A*	Malformation	Yes	*N/A*
Panas et al, 2005[108]	Europe	M	>12y	Complex	No	Yes
Panas et al, 2005[108]	Europe	M	>12y	Complex	No	Yes
Coban et al, 2010[109]	Asia	M	<1m	Malformation	No	No
Lim et al, 2010[110]	Asia	F	1-12m	Variant	No	No
Sartori et al, 2010[111]	Africa	M	1-12m	Malformation	No	No
Mademont-Soler et al, 2010[112]	Europe	F	>12y	Malformation	No	No
Stoodley et al, 2010[113]	North America	M	>12y	Malformation	*N/A*	*N/A*
Coulibaly et al, 2010[114]	Europe	M	1-12m	Malformation	Yes	No
Mytilinaios et al, 2010[115]	Europe	*N/A*	>12y	Variant	*N/A*	*N/A*
Unal et al, 2010[116]	Asia	M	1-12m	Malformation	No	No
Stevens et al, 2010[117]	South America	M	<1m	Malformation	No	Yes
Stevens et al, 2010[117]	South America	F	<1m	Malformation	No	Yes
Ramieri et al, 2011[118]	Europe	M	3-5y	Malformation	*N/A*	*N/A*
Marchal et al, 2011[119]	Europe	F	>12y	Malformation	*N/A*	*N/A*
Gverić-Ahmetasević et al, 2011[120]	Europe	F	1-12m	Malformation	No	No
Yahyaoui et al, 2011[121]	Europe	F	<1m	Variant	*N/A*	*N/A*
Tohyama et al, 2011[122]	Asia	F	1-12m	Malformation	No	No
Murphy-Ryan et al, 2011[123]	North America	M	1-12m	Malformation	No	No
Hu et al, 2011[124]	Asia	F	1-12m	Malformation	*N/A*	*N/A*
Turan et al, 2011[125]	Asia	M	>12y	Mega Cisterna Magna	*N/A*	*N/A*
Love et al, 2011[126]	Europe	M	3-5y	Malformation	No	No
Bunch et al, 2011[127]	North America	F	>12y	Variant	No	No
Tanoue et al, 2011[128]	Asia	F	13m-3y	Variant	*N/A*	*N/A*
Talamonti et al, 2011[129]	Africa	M	5-12y	Malformation	*N/A*	*N/A*
Hussain et al, 2011[130]	Asia	*N/A*	<1m	Malformation	*N/A*	*N/A*

Al-Achkar et al, 2011[131]	Asia	M	1-12m	Complex	No	No
Amin, 2012[132]	Asia	F	5-12y	Variant	*N/A*	*N/A*
Mancini et al, 2012[133]	South America	M	13m-3y	Variant	No	No
Manel et al, 2012[134]	Africa	F	<1m	Malformation	Yes	No
Aimua et al, 2012[135]	North America	F	>12y	Variant	*N/A*	*N/A*
Beby et al, 2012[136]	Europe	M	3-5y	Malformation	*N/A*	*N/A*
Kim et al, 2012[137]	Asia	F	1-12m	Variant	*N/A*	*N/A*
Cho et al, 2011[138]	Asia	F	1-12m	Malformation	*N/A*	*N/A*
Economou et al, 2012[139]	Europe	F	5-12y	Variant	No	No
Badakali et al, 2012[140]	Asia	F	1-12m	Malformation	Yes	*N/A*
Domínguez et al, 2012[141]	South America	F	>12y	Malformation	*N/A*	*N/A*
Kara et al, 2012[142]	Asia	M	>12y	Malformation	No	No
Kim et al, 2012[143]	Asia	M	5-12y	Malformation	No	No
Ryan et al, 2012[144]	Asia	F	>12y	Variant	*N/A*	*N/A*
Azukizawa et al, 2013[145]	Asia	F	1-12m	Malformation	No	No
Searson et al, 2013[146]	Europe	M	>12y	Variant	*N/A*	*N/A*
Kim et al, 2013[147]	Asia	M	>12y	Variant	No	No
Mandiwanza et al, 2013[148]	Europe	M	13m-3y	Malformation	*N/A*	*N/A*
Hackmann et al, 2013[149]	Europe	F	>12y	Malformation	No	No
Dhupar et al, 2012[150]	Asia	M	1-12m	Malformation	*N/A*	*N/A*
Pai et al, 2013[151]	Asia	F	>12y	Malformation	*N/A*	*N/A*
Zhang et al, 2013[152]	Asia	M	>12y	Malformation	No	No
Jang et al, 2013[153]	Asia	M	>12y	Malformation	*N/A*	*N/A*
Passalacqua et al, 2013[154]	South America	F	1-12m	Malformation	No	No
Zhou et al, 2013[155]	North America	F	>12y	Variant	*N/A*	*N/A*
John et al, 2013[156]	Asia	M	13m-3y	Variant	Yes	No
De Crecchio et al, 2013[157]	Europe	M	*N/A*	Complex	No	Yes
De Crecchio et al, 2013[157]	Europe	F	*N/A*	Complex	No	Yes
Guilherme et al, 2013[158]	South America	F	>12y	Variant	No	No
Bhattacharya et al, 2013[159]	Asia	M	3-5y	Variant	No	No
Sidana et, 2013[160]	Asia	M	>12y	Variant	*N/A*	*N/A*
Graf et al, 2013[161]	Europe	M	>12y	Variant	*N/A*	*N/A*

Mama et al, 2014[162]	Africa	F	>12y	Malformation	*N/A*	*N/A*
De Cock et al, 2014[163]	Europe	M	1-12m	Malformation	*N/A*	*N/A*
Rodríguez et al, 2014[164]	Europe	F	1-12m	Malformation	No	No
Na et al, 2014[165]	Asia	F	>12y	Malformation	*N/A*	*N/A*
Buonaquro et al, 2014[166]	Europe	F	>12y	Complex	No	No
Gathwala et al, 2014[167]	Asia	M	<1m	Variant	No	No
Pandurangi et al, 2014[168]	Asia	M	>12y	Complex	No	No
Pandurangi et al, 2014[168]	Asia	M	>12y	Complex	No	No

Abbreviations: DW: Dandy-Walker, F: Female, M: Male, N/A: Not Available, y: years old

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Table 2. Reported comorbidities under ICD 10 categorization

ICD10 Disease Category	**Comorbidities reported with Dandy Walker Syndrome (No of cases)**
II Neoplasm	nephroblastoma(Wilms tumour) (8) tongue hamartoma(4) rhabdomyosarcoma(2) meningeal melanocytoma (1) primary meningeal melanoma(1) intraventricular dermoid tumour (1) nasal glioma (1) hepatoblastoma(1) endobronchial carcinoid tumour(1) intracranial cavernous angioma(1) multiple tumoral lesions(1)
III Diseases of the blood and blood-forming organs and certain disorders involving the immune mechanism	Anaemia/thrombocytopenia(1) immunodeficiency(1)
IV Endocrine, nutritional and metabolic diseases	hirsutism(5) Hypothyroidism(2) molybdenium cofactor deficiency(2) hypoparathyroidism(1) precocious puberty(1) horseshoe adrenal gland(1) single-lobe thyroid(1) Hurler disease (mucopolysaccharidosis type I) (1) congenital disease of glycosylation(1) hyperinsulinism(1) nonketotic hyperglycinemia(1) *Carnitine palmitoyltransferase II* deficiency (CPT-II) (1) biotinidase deficiency(1)
V Mental and behavioural disorders	severe intellectual disability(10) bipolar disorder(5) psychosis(5) attention deficit hyperactivity disorder(4) mild mental abnormality(3) neurobehavioral abnormalities (auto and heteroaggressive disorder) (3) speech impairment(1) OCD(obsessive-compulsive disorder) (1)
VII Diseases of the eye and adnexa	cataracts(9) microphthalmia(6) microcornea/corneal opacity(5) myopia(5) optic nerve atrophy/dysplasia(5) chorioretinal atrophy/dysplasia(5) nystagmus(3) iris/optic nerve/optic disk coloboma(3) strabismus(2)

	anophthalmia(1) posterior embryotoxon(1)
-VIII Diseases of the ear and mastoid process	Congenital deafness/hearing loss(7) vertigo(1) laryngomalacia(1)
IX Diseases of the circulatory system	Patent ductus arteriosus(7) coarctation of aorta(6) ventricular septal defect(VSD) (6) atrial sepatl defect(ASD) (5) heart failure(5) pulmonary stenosis/hypoplasia(4) intracranial and extracranial vascular malformations(3) aortic valve anomay(2) tetralogy of Fallot(2) bilateral internal jugular vein occlusion(1) complex heart defect (ventricular and atrial septal defect, malrotation and interrupted aortic arch) (1) right sided aortic arch(1) total anomalous pulmonary venous return to the thoracic inferior vena cava, double outlet right ventricle and a type B interrupted aortic arch (1) engorgement of the main pulmonary artery(1)
XI Diseases of the digestive system	eosophageal atresia+ tracheoesophageal fistula/ bronchoesophageal fistula(1) Meckel's diverticulum(1) Hirschsprung disease(1) duodenal atresia(1) ectopic pancreas(1) multiple congenital liver masses(1) oesophageal diverticulum(1)
XII Diseases of the skin and subcutaneous tissue	Neurocutaneous melanosis/lipomatosis(11) hemangiomas(9) melanocytic nevi(6) skin and hair pigmentary problems(4) sparse hair/hair loss(3) cutis laxa(1) cutis aplasia(1) ichtyosis(1)
XIII Diseases of the musculoskeletal system and connective tissue	scoliosis/ kyphoscoliosis(4) joint laxity(4) Spinal Muscular Atrophy(SMA)(3) congenital arhtrogrypisis (contractures) (3) general muscular dystrophy(1) Mimicking Myasthenia Gravis(1)
XIV Diseases of the genitourinary system	hypoplastic genitalia(7) polycystic kidneys(4) dysplastic kidneys-kidney/renal agenesis/pelvic kidney (4) undescended testis/ cryptorchidism (4) hydronephrosis/ hydroureters(3) hypospadias(1)
XVII Congenital	PHACE syndrome (8)

malformations, deformations and chromosomal abnormalities	Trisomy 18 (Edwards syndrome)(4) Joubert syndrome(4) Oorthuys syndrome(3) oro-facio-digital syndrome (3) partial trisomy 3q(2) Coffin-Siris syndrome(2) Goldston syndrome(2) Kallmann's syndrome(2) Keratosis, Icthiosis, and Deafness (KID) Syndrome(2) trisomy 9q(1) partial trisomy of 8q(1) trisomy 22(1) 4q Deletion (1) 6p25 deletion(1) trisomy 21(1) translocations(1) Ritscher-Schinzel cranio-cerebello-cardiac syndrome(1) heterozygous loss of ZIC1 and ZIC4(1) Mayer-Rokitansky-Kuster-Hauser (MRKH) syndrome(1) bradycardia-tachycardia syndrome(1) multiple congenital anomalies/mental retardation (MCA/MR) syndrome(1) otopalatodigital syndrome type 2 (OPD2)(1) Neu-Laxova syndrome(1) Brown syndrome(1) Fowler syndrome(1) Moebius syndrome(1) Yunis-Varon syndrome(1) Ehlers-Danlos syndrome(1) Shah-Waardenburg syndrome(1) femoral-facial syndrome (FH-UFS)(1) Juberg-Hayward syndrome(1) Griscelli syndrome(1) Bobble-head doll syndrome (BHDS) (1) heterozygous missense mutation (D50N) in the GJB2 gene(1) oculocerebrocutaneous syndrome (OCCS) or Delleman–3C syndrome(1) microdeletion 3q syndrome(1)