Emanuel Syndrome (supernumerary der(22) syndrome): Brief summary of published cases

Year	References	Notable findings	Country reporting	# of cases
2022	Hao X, Wu J, Fu W, Zhang R, Zhong S, Deng Y, Zhu Y, Ye Y, Fang Q. Prenatal Diagnosis of Fetuses with Emanuel Syndrome: Results of Ultrasound Examination and Invasive Genetic Testing. Prenat Diagn. 2022 Jan 19. doi: 10.1002/pd.6098. https://obgyn.onlinelibrary.wiley.com/doi/10.1002/pd.6098	"All ES fetuses presented with multiple abnormalities by ultrasound examinations. Diaphragm hernia (3/6), Dandy-Walker complex (3/6), and kidney aplasia (3/6), were the most common ultrasound findings. Sonographic soft markers such as increased nuchal translucency, increased nuchal fold thickness"	China	13
2022	Adams, L. E., Chapman, A., Cormack, C. L., Campbell, K., Ebanks, A. H., Annibale, D. J., & Hollinger, L. E. (2022). Emanuel syndrome and congenital diaphragmatic hernia: A systematic review. *Journal of pediatric surgery*, *57*(9), 24–28. https://doi.org/10.1016/j.jpedsurg.2021.11.005	congenital diaphragmatic hernia (CDH)	USA	1

	https://pubmed.ncbi.nlm.nih.gov/34865829/			
2021	Hayakawa K, Kawase K, Fujimoto M, Nakamura Y, Saitoh S. Utility of breakpoint-specific nested polymerase chain reaction for the diagnosis of Emanuel syndrome. Pediatr Int. 2021 Dec;63(12):1534-1536. doi: 10.1111/ped.14644. Epub 2021 Aug 27. PMID: 34449117. https://pubmed.ncbi.nlm.nih.gov/34449117/	Not available.	Japan	
2020	Musa et al.. Diagnosis and Management of Genetic Derivation 22 and 11 Chromosome-Emanuel Syndrome in 10-Year Old Boy. Full text: https://www.fortunejournals.com/articles/diagnosis-and-management-of-genetic-derivation-22-and-11-chromosomeemanuel-syndrome-in-10year-old-boy.pdf	1 -10 year old boy Severe developmental delay, Hyperactivity, Hand-flapping, Flat feet, Loose joints, Unusually flexible fingers, Seizures Atrial septal defect, Recurrent pneumonia	Albania	1
2020	Kilic et al. Anesthetic management for tethered cord syndrome in a child with Emanuel syndrome. Full text: https://www.ejmanager.com/mnstemps/172/172-1593870579.pdf?t=1624	1-girl age 7 Growth delay, Aphasia, Mild pulmonary and tricuspid insufficiencies, Unilateral renal agenesis, Hip dislocation, Cleft palate Tethered cord syndrome.	Turkey	1

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2020	Lou et al. Non-invasive prenatal screening for Emanuel syndrome. Full text: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC7057502/	both cases mother was t(11;22) carrier. Dandy Walker malformation in 1 case	China	2
2020	Yik et al.. A case study of distinctive phenotypes arising from Emanuel syndrome in two karyotypically identical patients. Full text: https://medic.upm.edu.my/upload/dokumen/2020051111452012_MJMHS_0463_11_5_2020.pdf (Reviewer disputes some of the comments within this article)	1 - female Low set ears with skin tags, Micrognathia Mother is carrier 2 - girl Micrognathia, Low set ears, Bilateral dislocatable radius and ulna, Limited hip adduction, High arched palate, Microcephaly Bilateral preauricular pits, Mother is carrier	Malaysia	2
2020	Edoardo et al. Early detection of Emanuel syndrome: a case report. Abstract: https://www.degruyter.com/document/doi/10.1515/crpm-2020-0049/html	No abstract available	Switzerland	1
2019	Kamath et al. The Constitutional balanced translocation t(11;22)(q23;q11.2)-An Indian account. Full text: https://www.jcdr.net/articles/PDF/12438/36950_CE[Ra1]_F(SL)_PF1(AGAK)_PFA(AK)_PB(AG_SHU)_PN(SL).pdf	1-girl age 6 Hypotonia, Seizures, Developmental delay Cleft palate, Ventricular septal defect, Auditory and visual insufficiency, Malrotation of gut Mother t(11;22) carrier 2-boy age 8 months Developmental delay, Dandy-Walker malformation, Atrial septal defect, Abnormal testes, Microtia, Hearing impairment	India	3

		3-girl age 3 Developmental delay, Microcephaly Seizures, Atrial septal defect		
2019	Puranik & Katechia. Oral and dental findings in Emanuel syndrome. Abstract: https://onlinelibrary.wiley.com/doi/abs/10.1111/ipd.12502	1-boy - age followed from 3-14 Mother carrier Microcephaly, Micrognathia, Preauricular pits Bilateral hearing loss, Microtia, Recurrent ear infections, Hypotonia, Non-verbal, Seizures Self-injurious behaviour, Delayed puberty Atrial and ventricular septal defects, Patent ductus arteriosus, Micropenis, Hypospadias Hypoplastic kidney, Hernia (repaired at 7) Decreased immunoglobulin level, Gastroesophageal reflux, G-tube fed Hip dislocation, Club foot, Supernumerary pairs of ribs, Clinodactyly, Hyperextensibility of joints	USA	1
2019	Xie & Cardenas. Neuroimaging findings in Emanuel Syndrome. Full text: https://pdfs.semanticscholar.org/94aa/0c5a3e027c52a3acef6ea5315a56b3cdaa99.pdf?_ga=2.140527922.1203572888.1623707750-175906158.1623707750	1-boy Mother had balanced t(11;22) Hypotonia, Cleft palate, Micrognathia Retrognathia, Microcephaly, External auditory canal atresia, Cerebral volume loss, Recurrent ear infections, Bilateral sensorineural hearing loss, Dysplasia of the bilateral middle ear ossicles	USA	1
2019	Karempelis et al. Associated syndromes in patients with Pierre Robin sequence. https://www.sciencedirect.com/science/article/abs/pii/S0165587619305956?via%3Dihub	Pierre Robin sequence in two cases	USA	2

2019	Antonenko et al. A case of Emanuel syndrome on a newborn girl with congenital heart defect. Full text - in RUSSIAN - information obtained by using google translate to obtain data: https://www.medgen-journal.ru/jour/article/view/721/446	Father t(11;22) carrier Intestinal obstruction at 28 days, Atrial septal defect, Interventricular septal defect, Patent ductus arteriosus, Retinal angiopathy, Hypertonia, preauricular tags	Russia	1
2019	Li et al. Prenatal diagnosis of a fetus with trisomies of 11q23.3q25 and 22q11.1q11.21. Article in Chinese. Abstract: https://pubmed.ncbi.nlm.nih.gov/31055824/	1 - boy Dandy-Walker syndrome, growth restriction, and right-heart system dysplasia. Father t(11;22) Carrier	China	1
2019	Zienkiewicz. Emanuel syndrome. The case study. https://edu.eacd.org/node/620	7 year old girl Walked at age 3 alone. Atrial septal defect, Hypotonia, Bilateral hearing loss, cleft palate Hypoplasia of one kidney	Poland	1
2018	Saxena et al. Phenotypic characterization of derivative 22 syndrome: case series and review. Full text: https://www.ias.ac.in/article/fulltext/jgen/097/01/0205-0211	1-boy - 22 months old Delayed milestones, Left preauricular pit Mother carrier of t(11;22) 2- female - 10 months Craniostenosis, Right coronal suture fusion Mother carrier of t(11;22) 3- male - 8 months Feeding difficulties, Developmental delay Ear deformity, Recurrent respiratory tract infections 4-girl 6 years old	India	4

		Cleft palate, Seizure episode, Atrial septal defect, Preauricular skin tags and pit Mother t(11;22) carrier		
2018	Kankante et al. Emanuel syndrome: A case report. Full text: http://www.vimshsj.edu.in/index.php/main/article/view/104/80	Male Microcephaly, Unilateral right sided microtia Large preauricular skin tags and pit, Small penis, Micrognathia, High arched palate Moderately large ventricular septal defect Small patent ductus arteriosus, Hypotonia Developmental delays, Growth parameters below 3rd percentile	India	1
2018	Saffren et al. Ocular manifestations of Emanuel syndrome. Abstract: https://onlinelibrary.wiley.com/doi/abs/10.1002/ajmg.a.40361	36 year old male Cataract left eye at age 31 Glaucoma in right eye at 32 Glaucoma in left eye at 35. Pierre Robin sequence, Recurrent ear infections, Inguinal hernia, Cleft palate, Multiple bilateral ear tubes, No light perception in left eye - left pupil unresponsive to light, Marked left nuclear sclerosis, High myopia See article for full details on ocular findings	USA	1
2018	Shenoy et al. Chromosomal abnormalities in syndromic orofacial clefts: Report of three children Full text: Chromosomal Abnormalities in Syndromic Orofacial Clefts: Report of Three Children - PMC	Male - 10 months Developmental delay, Seizures, Microcephaly Hypotonia, Cleft palate, Micrognathia,Thinning of corpus callosum Mother 11/22 carrier	India	1
2018	Soto-Brambila et al. Emanuel	1 case - 3 year old girl	Mexico	1

	Syndrome: First case reported in Mexico. Free full text: https://produccioncientificaluz.org/index.php/investigacion/article/view/29078/29798	Delayed development, Bilateral hip dysplasia Cleft palate, Patent ductus arteriosus Multiple ear and respiratory infections 1 episode seizure , Micrognathia Mild bilateral sensorineural hearing loss Mother is translocation carrier		
2017	Liehr et al. Next generation phenotyping in Emanuel and Pallister-Killan syndrome using computer-aided facial dysmorphology analysis of 2D photos. Abstract: https://onlinelibrary.wiley.com/doi/abs/10.1111/cge.13087			
2017	Kashiwagi et al. Efficacy of intravenous immunoglobulin in a case of Emanuel syndrome. Full text: https://www.scirp.org/pdf/OJPed_2017030915523960.pdf	Male Ear pits and tags, Pierre Robin sequence Micrognathia, Severe hearing loss excess nuchal skin, Abnormalities of ear shape, size, position, Small penis, Atrial septal defect Ventricular septal defect, Pulmonary stenosis Hypoxia-related brain encephalopathy Gastroesophageal reflux, Cleft palate Seizures. Frequent infectious diseases Low IgG level	Japan	1
2017	Zhao et al. Genetic diagnosis and follow up of a fetus with Emanuel syndrome. Article in Chinese.https://pubmed.ncbi.nlm.nih.gov/28981939/	Dandy Walker malformation Mother carrier of t(11;22)	Chinese	1
2017	Luo et al. A clinical and molecular	1-boy age 2	Chinese	1

	analysis of a patient with Emanuel syndrome. Full text: https://www.spandidos-publications.com/10.3892/mmr.2017.6107?text=fulltext	Atrial septal defect, Thymic dysplasia Cleft lip and palate Genital malformations (micropenis)		
2016	Zou et al. A rare case of trisomy 11q23.3-11q25 and trisomy 22q11.1-22q11.21 https://www.geneticsmr.com/sites/default/files/articles/year2016/vol15-2/pdf/gmr8140_1.pdf	Developmental delay, speech impairment, hypotonia, microcephaly,micrognathia, high arched palate, agenesis of the corpus callosum, cerebellar vermis hypoplasia	China	1
2016	Tsukamoto et al. Anesthetic Management of a Patient With Emanuel Syndrome Full text: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC5157147/	tracheomalacia, micrognathia, atrial septal defect (ASD), mild pulmonary valve stenosis (PS), cleft palate, preauricular tags, severe hearing loss, and microtia	Japan	1
2016	Vahidi Mehrjardi et al. Newborn with supernumerary marker chromosome derived from chromosomes 11 and 22 - a case report. Full text: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4851753/	1-girl Intrauterine growth retardation, Dysplastic ears Skin tag. Cleft palate. Atrial septal defect Systolic ejection of the left ventricle Hypoplasia of the corpus callosum and septum pellucidum	Iran	1
2016	Deepika et al. Prenatal assessment of three rare syndromes from Telangana Region by 3D/4D Sonography. Full text: https://www.longdom.org/open-access/prenatal-assessment-of-three-rare-syndromes-from-telangana-region-by-3d4d-sonography-2157-7412-10003	1-girl Intrauterine growth retardation, Dysplastic ears Congenital heart defects (small VSD and ASD along with moderate PDA and PDH) Imperforate anus, Sacral dimple Developed acute bilirubin encephalopathy and passed at 5 months of age	India	1

	09.pdf			
2015	Kapoor. Emanuel syndrome: a rare disorder that is often confused with Kabuki syndrome. Full text: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4489075/	Commentary	USA	
2015	Nishinarita et al. Anesthetic management of pediatric patients with Emanuel syndrome. Abstract: https://link.springer.com/article/10.1007/s00540-014-1955-y	1-boy Malformed ears with pits, Micrognathia Stenosis of the proctodeum, Membranous anal atresia, Sacral dimple, Cleft palate Single kidney,Atrial septal defect Patent ductus arteriosus 2 - girl Bilateral microtia, Atresia of the right auditory meatus, Cleft palate, Left macrostomia Atrial septal defect with mild pulmonary artery stenosis, Laryngomalacia, Glossoptosis	Japan	2
2015	Jancevska et al. Emanuel syndrome (ES): New case report and review of the literature. Full text: https://www.researchgate.net/publication/282548777_Emanuel_Syndrome_Es_New_Case-Report_and_Review_of_the_Literature_Emanuel_Sindrom_Es_Prezentacija_Na_Nov_Slucaj_I_Pregled_Na_Literaturata	1-boy - age 2 Preauricular pit Micrognathia Small penis Significant delays ambulatory	Macedonia	1
2015	Ikbal Atli et al. A case with Emanuel syndrome: Extra derivative 22 chromosome inherited from the mother.	1 - boy-3 years old Cleft palate, Micrognathia, Undescended testes Inguinal hernia, Atrial septal defect, Hypotonia, White matter anomalies, Skeletal anomalies,	Turkey	1

	https://www.researchgate.net/publication/299853842_A_Case_with_Emanuel_Syndrome_Extra_Derivative_22_Chromosome_Inherited_from_the_Mother	Cerebral atrophy, Scoliosis & kyphosis, Glaucoma, Gallstones, Feeding issues recurrent ear and respiratory infections Mother carrier of t(11;22)		
2015	Zou et al. A rare case of trisomy 22q13.3-11q25 and trisomy 22q11.1-22q11.21. Full text: https://www.geneticsmr.com/sites/default/files/articles/year2016/vol15-2/pdf/gmr8140_1.pdf	1-girl age 6 Mother carrier t(11;22) Brain malformations, Microcephaly, Hypotonia Micrognathia, High arched palate, Agenesis of the corpus callosum, Cerebellar vermis hypoplasia, Simple cerebral gyration	China	1
2014	Zafar, Chaudari & Crossland. Emanuel syndrome with unique cardiac defects: a case series. Full text: https://www.infantjournal.co.uk/pdf/inf_057_dia.pdf	1 - female Ear tags, Skin tags, Small chin, Microcephaly Mild hypotonia, Talipes of the feet, Microphthalmia, Mild renal failure Mother is 11/22 carrier Case 2 - male Small chin, Cleft palate, Growth restriction Interruption of the aortic arch, Mitral hypoplasia Small left ventricle, Aortic hypoplasia Hypoplastic arch, Died of heart related issues within 24 hours of birth.	UK	2
2014	Dudarewicz et al. Emanuel syndrome. The Fetus. https://thefetus.net/content/emanuel-syndrome/	hypoplasia of the lower cerebellar vermis, bilateral markedly hypoplastic kidneys, micrognathia, small stomach with polyhydramnios, borderline IUGR, and later rocker-bottom feet, annular pancreas, mother carrier.	Poland/Germany	1
2014	Ohye et al.. Prevalence of Emanuel	Obtained data to assess prevalence only	Japan	36

	syndrome: Theoretical frequency and surveillance result. Full text: https://onlinelibrary.wiley.com/doi/epdf/10.1111/ped.12437			
2013	Fokstuen et al. Contamination of amniotic fluid with maternal balanced t(11;22) translocation cells. Abstract: https://onlinelibrary.wiley.com/doi/abs/10.1002/ajmg.a.35774	Fetal death 17 weeks Microcephaly, Right sided diaphragmatic hernia Only one ovary	Switzerland	1
2013	Choudhary et al. Derivative 11;22 (Emanuel) syndrome: A case report and a review. Full text: https://www.hindawi.com/journals/cripe/2013/237935/	1-male High arched palate, Micrognathia, Ventricular septal defect, Small penis, Missing right kidney Mild hearing loss, Significant hypotonia Mother carrier of t(11;22)	India	1
2013	Kee et al. Differential outcomes in an extended family with constitutional t(11;22)(q23.3;q11.2). Full text: https://www.researchgate.net/publication/306268846_Differential_outcomes_in_an_extended_family_with_constitutional_t1122q233q112	Male child Mother carrier	Singapore	1
2013	Dudarwicz et al. Emanuel Syndrome. Full text: https://thefetus.net/content/emanuel-syndrome/	hypoplasia of the lower cerebellar vermis, bilateral markedly hypoplastic kidneys, micrognathia, small stomach with polyhydramnios, borderline IUGR, and later rocker-bottom feet, annular pancreas Moher t(11;22) carrier	Germany	1
2012	Glaser et al. Lipodermoid in a patient with Emanuel syndrome. Abstract https://www.sciencedirect.com/scienc	1-boy 8 month Bilateral absence of ear canals, Microtia Multiple auriculaire pits, Retrognathia	USA	1

	e/article/abs/pii/S1091853113000645	Mandibulaire hypoplasia, Right facial hemiparesis, Conductive hearing loss Patent ductus arteriosus, ventricular septal defect, Conjunctival lipodermoid in the right eye		
2012	Kim et al. A case with Emanuel syndrome resulting from a maternal balanced translocation. Full text: https://www.koreascience.or.kr/article/JAKO201225038987926.pdf	1- girl mother t(11;22) carrier and one sister carrier Cleft palate, Low set ears, Micrognathia Small left kidney, Vesicoureteral reflex grade II Atrial septal defect, Sensorineural hearing loss Repeated aspiration pneumonia, Severe velopharyngeal incoordination upon swallow study, Seizures, Hypotonia, Diffuse brain atrophy and thinning of corpus callosum Severe global developmental delay	Korea	1
2012	Walfisch et al. Prenatal screening characteristics in Emanuel syndrome: a case series and review of the literature. Abstract: https://pubmed.ncbi.nlm.nih.gov/22434056/	Data only available for three/five prenatally diagnosed cases 1 - female Increased nuchal fold, Posterior fossa abnormalities, Dandy-walker malformation and mild ventriculomegaly, Left diaphragmatic hernia, Sacral splaying of the spine, Echogenic intra-cardiac focus, Echogenic bowel, Rocker bottom feet deformities, Growth restriction 2- female Growth restriction,Aortic coarctation Pelvicalyceal dilation 3 - male Growth restriction, Posterior fossa cyst inferior	Canada	5

		to the cerebellum consistent with Dandy walker complex, cleft palate, Micrognathia, Abnormal ears, Duodenal atresia, Abnormal male genitalia		
2010	Gordillo González G, Rojas A, Zarante I, Gómez A, Moreno OM, Osorio G. Emanuel syndrome or supernumerary derived chromosome 22 syndrome. Iatreia [Internet]. 2010 Nov 28 [cited 2021 Jun 29]; 23 (4-S): S-78. Full text: https://revistas.udea.edu.co/index.php/iatreia/article/view/8227	1-boy 3 months Microcephaly Micropenis Unilateral agenesis and inguinal hernia Ambiguous genitalia Mother t(11;22) carrier	Columbia	1
2010	Garcia-Vielma C, Emanuel syndrome (supernumerary derivative 22), the result of a maternal translocation. A case report. Journal of the Association of Genetic Technologists 36(4):189-93 Catalina García-Vielma, Rosa Maria de la Rosa-Alvarado, Karem Nieto-Martinez, Elva Irene Cortes-Gutierrez https://www.researchgate.net/publication/49675804_Emanuel_syndrome_supernumerary_derivative_22_the_result_of_a_maternal_translocation_A_case_report	1 girl age 6 Anorectal anomaly Hip dysplasia Microtia Cyst on kidney	Mexico	1

2010	Soo Jun et al. A case of supernumerary derivative (22) syndrome resulting from a paternal balanced translocation. KOREAN. Google translate used to obtain data. Full text: https://neo-med.org/upload/pdf/J%20Korean%20Soc%20Neonatol._17_1_127_131.pdf	1-girl - 1 day old Hypotonia, Low set ears with pit, Micrognathia High arched palate, Bilateral hip subluxation Bilateral shoulder joint subluxation, Atrial septal defect, Bilateral hearing loss, Kidney malformation Father t(11;22) carrier	Korea	1
2010	[Communication support for children with Emanuel Syndrome] translated title. Google translate used to obtain data.Full text Article in Japanese: https://core.ac.uk/download/pdf/153445718.pdf	1-boy age 7 Severe mental delay, Microcephaly, Delayed growth, Atrial septal defect, Hearing loss Visual impairment, Prone to infectious diseases Seizures	Japan	1
2010	Rosa et al. Phenotypical variability in supernumerary chromosome der(22)t(11;22) syndrome (Emanuel syndrome). Full text - article in Portuguese. Google translate used to obtain data. https://www.scielo.br/j/rpp/a/jtTRxFjvJTn3fNQMHM5Wgxv/?lang=pt&format=pdf	1-girl age 2 years Hypotonia, Delayed development Stereotypical movements/autistic behaviour Microcephaly, Ear pits and tags, Imperforate anus, Cerebral hypoplasia, Pulmonary valve stenosis, Laryngotracheomalacia, Cleft palate Multiple respiratory infections Mother carrier 2 - boy age 6 months Hypotonia, Growth deficit, Microcephaly Hemifacial microsomia, Cleft palate, Microtia Ear tags, Proximally implanted thumbs Pulmonary valve stenosis, Interatrial and interventricular communication, patent ductus arteriosus, left superior vena cava Oculo-auriculo-vertebral spectrum (OAVS)	Brazil	2

		Father carrier		
2010	Fabiano et al. Phenotypic variability in chromosome syndrome supernumerary der(22) t(11;22) (Emanuel syndrome). Full text in Portuguese, google translate used to obtain data: https://www.scielo.br/j/rpp/a/jtTRxFjvJTn3fNQMHM5Wgxv/?lang=pt&format=pdf	1 - girl - age 5 Hypotonia, Microcephaly, Ear pits and tags Imperforate anus, Cerebral hypoplasia Pulmonary valve stenosis, Laryngomalacia Cleft palate Mother t(11;22) carrier 2- boy - 6 months Hypotonia, Growth deficiency, Microcephaly Hemifacial microsomia, Cleft palate, Micrognathia, Preauricular tags, Proximally implanted thumbs, Pulmonary valve stenosis, Ventricular septal defect, Persistent ductus arteriosus, Left superior vena cava Father t(11;22) carrier	Brazil	2
2010	Kazendi et al. Emanuel syndrome. Full text (in Turkish) Google translate used to obtain data: http://egetipdergisi.com.tr/tr/download/article-file/350438	1-boy Skin tags on ear and cheek, Increased nuchal fold, Microcephaly, Ventriculomegaly Micrognathia, Cleft palate, Renal anomalies Genitourinary system anomalies, Diaphragmatic hernia, Complete endocardial cushion defect, Truncus arteriosus Pregnancy terminated at 20 weeks. Mother t(11;22) carrier	Turkey	1
2009	Carter et al. Phenotypic delineation of Emanuel syndrome (Supernumerary Derivative 22 syndrome): Clinical features of 63 individuals. Full text: Phenotypic delineation of Emanuel syndrome	63 cases from USA, Canada, UK, Australia, France, Italy, Norway, SPain, Chile 95% mother was carrier, Only 2 father as carrier, One adoptive parent, 28 males, 35 females - Age ranges 9 months to 33 years	International	63

	(Supernumerary Derivative 22 syndrome): Clinical features of 63 individuals. Open access - full text: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2733334/	Reports on diagnosis and counselling, carrier status of parents, pregnancy and neonatal period, Congenital anomalies, dysmorphism, medical issues, growth and puberty, neurological issues, development and behaviour		
2009	Toyoshima, et al. Vertebral fusion in a patient with supernumerary-der(22)t(11;22) syndrome. Abstract: https://onlinelibrary.wiley.com/doi/10.1002/ajmg.a.32762	1-girl T1-T2 vertebral fusion (congenital), Anal atresia Atrial septal defect, Cleft palate, Bilateral preauricular tags, Feeding difficulties, Bilateral congenital hip dislocation, Walked independently at age 4, Seizures onset at age 16, Kyphoscoliosis, Sprengel deformity Mixed deafness, Strabismus Mother carrier	Japan	1
2009	Gremeau, et al. Congenital diaphragmatic hernia and genital anomalies. Abstract: https://pubmed.ncbi.nlm.nih.gov/19434659/	1-boy Left-sided congenital diaphragmatic hernia (agenesis left diaphragm), Micropenis Left preauricular tag, 13 pairs of ribs Hypoplastic posterior corpus callosum Persistent pulmonary hypertension Intrahepatic cholestasis, Necrotizing enterocolitis, sepsis Passed day 23 from multiple organ dysfunction	France	1
2008	Afroze et al. Supernumerary derivative (22) syndrome resulting from a maternal balanced translocation. Full text http://www.smj.org.sg/sites/default/files/4912/4912cr11.pdf	All growth parameters below 3rd percentile. Bilateral microtia, Supra-auricular skin tags Skin pits, Cleft palate, Micrognathia Ventricular septal defect, Patent ductus arteriosus, Significant hypotonia Mother t(11;22) carrier	Malaysia	1

2008	Boroova et al. Constitutional translocation t(11;22) in Slovak Romanies from the Presov Region (Slovakia). Abstract: http://www.krepublishers.com/02-Journals/T-Anth/Anth-10-0-000-08-Web/Anth-10-1-000-08-Abst-PDF/Anth-10-1-001-08-377-Boro%C5%88ov%C3%A1-I/Anth-10-1-001-08-377-Boroova-I-Ab.pdf	1 - male Mother carrier 2 - female Father carrier 3 - female Passed after 48 hours following birth Father carrier	Slovakia	3
2008	Allotey et al. Congenital bile duct anomalies (biliary atresia) and chromosome 22 aneuploidy. Abstract: https://www.jpedsurg.org/article/S0022-3468(08)00430-2/pdf	Biliary atresia	United Kingdom	2
2007	Fenerci et al. Supernumerary chromosome der(22)t(11;22): Emanuel syndrome associates with novel features. Abstract: https://pubmed.ncbi.nlm.nih.gov/18286821/	Boy - 9 years old Joint hyperextensibility, elbow and wrist joints Left liver lobe agenesis, Left sided malposition of the gallbladder, Pancreas hypoplasia Genital anomalies, Developmental delay Seizures, Autistic behaviour (absent eye contact and stereotypical hand movements) Body size below 3rd percentile for age Mild-moderate hypotonia, Limited vocabulary capacity, High arched narrow palate Teeth irregularities with multiple caries Hypoplastic scrotum, Bilateral cryptorchidism Clinodactyly of the fifth fingers Dandy-Walker malformation Dilatation of the cerebral ventricular system	Turkey	1

		Mother and maternal grandmother carriers of 11/22		
2007	Prieto et al. Phenotypic expansion of the supernumerary derivative (22) chromosome syndrome: VACTERL and Hirschprung's disease. Abstract: https://www.jpedsurg.org/article/S0022-3468(07)00510-6/fulltext	2 males 1 with VACTERL 1 with Hirschsprung's disease	USA	2
2006	Chen et al. Prenatal diagnosis of the supernumerary der(22)t(11;22) syndrome associated with abnormal sonographic findings. Abstract: https://obgyn.onlinelibrary.wiley.com/doi/abs/10.1002/%28SICI%291097-0223%28199612%2916%3A12%3C1137%3A%3AAID-PD979%3E3.0.CO%3B2-K	1-male fetus Microcephaly Micrognathia Ear anomalies Small penis	Taiwan	1
2006	Balci et al. Partial trisomy (11;22) syndrome with manifestations of Goldenhar sequence due to maternal balanced t(11;22). Abstract: https://pubmed.ncbi.nlm.nih.gov/17100195/	1-girl age 15 Cleft palate, micrognathia, Hemifacial microsomia, Hypoplasia of the external auditory canal, Bilateral preauricular skin tags and dimple, Microphthalmia, Umbilical and inguinal hernia, Growth below 3rd centile, Microcephaly Hypotonia, Kyphoscoliosis, Right renal agenesis, Hypoplasia of right external auditory canal and right middle ear and dysplasia of middle ear bones.Corpus callosum hypoplasia	Turkey	1
2006	Guven et al. Prenatal diagnosis of a case with Emanuel syndrome	1-female fetus Partial cerebellar vermian agenesis and	Turkey	1

	(supernumerary der(22) syndrome). FUll text: https://obgyn.onlinelibrary.wiley.com/doi/epdf/10.1002/uog.3429	posterior fossa dilatation in cranium with the diagnosis of Dandy-Walker malformation. Pulmonary stenosis, ventricular septal defect, posterior position of the pulmonary artery, diagnosis of Tetralogy of Fallot, Low set ears Cleft palate, Micrognathia, Microcephaly Ear abnormalities		
2005	Drum et al. Anaesthesia in a patient with chromosome 11;22 translocation: a case report and literature review. Abstract: https://onlinelibrary.wiley.com/doi/abs/10.1111/j.1460-9592.2005.01561.x	1-girl age 8 Large epiglottis, Hydrocephalus Heart defect, Imperforate anus Cleft palate	USA	1
2004	Imakata et al. Trisomy 11/22 diagnosed by spectral karyotyping (SKY). https://pubmed.ncbi.nlm.nih.gov/15517836/	1-male Microcephaly, Uvular hypoplasia Ear deformity, cryptorchidism	Japan	1
2003	Werding et al. Partial trisomy 22 resulting from rearrangements between chromosomes 11/22 and 16/22: A report of two cases. Abstract: https://onlinelibrary.wiley.com/doi/abs/10.1111/j.1651-2227.2003.tb02550.x?sid=nlm%3Apubmed	1 - Hypotonic, Low set ears and preauricular appendages, Cryptorchidism, Micropenis Dandy-Walker malformation, Smaller left kidney Aortic stenosis and pulmonary stenosis Mother t(11;22) carrier	Germany	1
2003	Chetcuti-Ganado & Grech. Complex congenital cardiac disease in a patient with partial trisomy for the long arms of chromosomes 11 and 22. Full text:	1-male Right preauricular pit, Coarctation of the aorta Atrioventricular septal defect with common atrioventricular junction, separate valvar orifices for the right and left ventricles, and exclusively	Malta	1

	https://www.researchgate.net/publication/8942681_Complex_congenital_cardiac_disease_in_a_patient_with_partial_trisomy_for_the_long_arms_of_chromosomes_11_and_22	atrial shunting. Arterial duct patent and supracardiac total anomalous pulmonary venous connection. Single kidney Tracheomalacia Mother t(11;22) carrier		
2003	Rassu et al. Der(11)t(11/22) resulting from a balanced reciprocal subtelomeric translocation in a Sardinian family. 4th European Cytogenetics Conference, 2003 Italy.	1 Microcephaly, Cleft palate Undescended testes, Low set ears	Sardinia	1
2003	Hee Yeon Woo, et al. Marker chromosomes in Korean patients: Incidence, identification and diagnostic approach. Full text: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3055124/	1-girl (case 5 of report) Intrauterine growth retardation, hypotonia, preauricular tag, ptosis of left eye	Korea	1
2003	V. Ivanovic & S. Grkovic. Unbalanced 11;22 translocation - case report. Abstract - 4th European Cytogenetics Conference 2003 Italy.	1-boy 18 months Anal stenosis, Microcephaly, Dysmorphic - malformed ears, preauricular pits, High palate Hypotonia, Heart defect, Hip dislocation Delayed appearance of teeth, Seizures Frequent respiratory infections	Serbia	1
2003	Cocchi, R. Neurologist. Twenty-seven months of antistress drug therapy in a girl with partial trisomy 22. Originally posted to personal website but no longer available online.	1 - girl - 16 years old Kyphosis, cleft palate, Heart defect (atrial)	Italy	1
2003	Vigliano et al. Hippocampal	10 year old boy	Italy	1

	malrotation in a supernumerary der(22) syndrome and epilepsy: a case report. Full text: https://tspace.library.utoronto.ca/bitstream/1807/1541/1/pn03008.pdf	Microcephaly, Strabismus, Micrognathia Left preauricular tag, Abnormal conformation of left ear, Renal ectopia, Cryptorchidism Congenital hip dysplasia, Visual defect Bilateral neurosensorial and conductive hearing loss, Seizures, Hypoplasia of the corpus callosum, Crawled 18 months, Walked 4-6 years, A few spoken words Mother carrier		
2002	Hou. Supernumerary chromosome marker der(22)t(11;22) resulting from a maternal balanced translocation. Full text: http://cgmj.cgu.edu.tw/2601/260107.pdf	1 - boy Small, hypoplastic kidneys - acute renal failure at 1 month, All growth below 3rd percentile Micrognathia, High arched palate, Underdeveloped ears, Preauricular pits Patent ductus arteriosus, Atrial septal defect, pulmonary valve stenosis, Brain anomalies Sensorineural hearing impairment, Micropenis Right cryptorchidism, Bilateral hydrocele Microcephaly, Failure to thrive Mother t(1122)carrier	Taiwan	1
2002	Kulharya et al. Unusual mosaic karyotype resulting from adjacent 1 segregation of t(11;22): Importance of performing skin fibroblast karyotype in patients with unexplained multiple congenital anomalies. Abstract: https://pubmed.ncbi.nlm.nih.gov/12457409/	1-male age 26 Mosaic Karyotype - 46,XY,der(22)t(11;22)(q23.3;q11.2)[7]/46,XY[143] Pierre Robin sequence, Micrognathia Mother t(11;22)	USA	1
1998	de Lonlay-Debeney et al. Ebstein	1- girl - died following birth	France	1

	anomaly associated with rearrangements of chromosome region 11q. ABstract: https://onlinelibrary.wiley.com/doi/abs/10.1002/(SICI)1096-8628(19981102)80:2%3C157::AID-AJMG12%3E3.0.CO;2-U	Ebstein anomaly (heart condition) Pierre Robin sequence Bilateral renal hypoplasia Mother t(11;22) carrier		
1997	Kadir et al. Prenatal diagnosis of supernumerary chromosome derivative (22) due to maternal balanced translocation in association with diaphragmatic hernia: A case report. Abstract: https://obgyn.onlinelibrary.wiley.com/doi/abs/10.1002/%28SICI%291097-0223%28199708%2917%3A8%3C761%3A%3AAID-PD121%3E3.0.CO%3B2-A	1-girl Diaphragmatic hernia Mild ventriculomegaly, dilatation of the third ventricle Mother t(11;22) carrier	UK	1
1996	Petkovic et al. Unusual segregation of t(11;22) resulting from crossing-over followed by 3:1 disjunction at meiosis I. Abstract: https://onlinelibrary.wiley.com/doi/abs/10.1111/j.1399-0004.1996.tb02725.x	1-girl age 4 Micrognathia, Cleft palate, Hypotonia, Preauricular pits/tags, Dislocated hips Strabismus, Dysplastic ears, Atrial septal defect Seizures, Recurrent infections Father t(11;22) carrier	Italy	1
1996	Pallotta et al. Cerebral defects confirm midline developmental field disturbances in supernumerary der(22),t(11;22) syndrome. Abstract: https://onlinelibrary.wiley.com/doi/abs/10.1111/j.1399-0004.1996.tb02398.x	1-girl Strabismus, Cleft palate, Micrognathia Dysplastic ears with right preauricular tag Telethelia, Hypotonia, Cardiac: patent Botallo duct and foramen ovale, anomalous venous pulmonary drainage and wide interatrial defect with pulmonary hypertension, Cerebral - bilateral ventricular dilatation with	Italy	1

		enlarged cisterna magna, posterior hypoplasia of corpus callosum, hyperechogenicity at the basal nuclei, microcephaly, hydrocephaly Delayed psychomotor development Mother t(11;22) carrier		
1996	Dawson et al. Der(22)t(11;22) resulting from a paternal de novo translocation, adjacent 1 segregation, and maternal heterodisomy of chromosome 22. Full text: https://jmg.bmj.com/content/jmedgenet/33/11/952.full.pdf	1-girl - age 8 years Preauricular pits and ear tags, High arched palate, Micrognathia, Flexor contractures of the elbows and knees, Dislocated right hip Rocker bottom feet, Complex cardiac anomalies incl. Tetralogy of Fallot, AV canal, large PDA. Father t(11;22) carrier	Canada	1
1996	Stankiewicz, P., Korniszewski, L., Bocian, E., & Stańczak, H. (1996). Marker chromosomowy jako produkt rodzinnej translockacji (11;22) rozpoznanej metodami cytogenetyki molekularnej [Marker chromosomes as a product of familial translocation (11;22) identified with molecular cytogenetic methods]. Pediatria polska, 71(3), 241–245. https://pubmed.ncbi.nlm.nih.gov/8966096/	1 - male Facial dysmorphology, hypotonia, heart failure, cryptorchism	Poland	1
1996	Chen et al. Prenatal diagnosis of supernumerary der(22)t(11;22) associated with the Dandy-Walker malformation in a fetus. Abstract: https://obgyn.onlinelibrary.wiley.com/doi/abs/10.1002/%28SICI%291097-0223%28199612%2916%3A12%3C11	1-male fetus Low-set malformed ears with preauricular tags High arched palate, Micrognathia Cerebellar dysgenesis, Retrocerebellar cyst Dandy-Walker malformation Mother t(11;22) carrier	Taiwan	1

	37%3A%3AAID-PD979%3E3.0.CO%3B2-K			
1995	Blancato et al. Prenatal diagnosis of partial trisomy through in Situ hybridization on amniocytes with whole chromosome and centromere-specific DNA probes: A case report. Abstract: https://pubmed.ncbi.nlm.nih.gov/7473445/	1-male Large left fetal congenital diaphragmatic hernia Heart in the right chest, and small right pleural effusion, stomach and bowel in the left side of the chest, Malrotation and displacement of the portal sinus, Hypoplastic genitalia, Small anus Mother t(11;22) carrier		1
1992	Tachdjian et al. Unbalanced karyotype due to adjacent 1 segregation of t(11;22)(q23.2q13.2). Abstract: https://pubmed.ncbi.nlm.nih.gov/1296521/	1-female Craniofacial dysmorphism, High arched palate Strabismus, Microretrognathia, Low set ears Sacral dimple, Dislocation of the right hip Supernumerary rib, hypotonia Mother t(11;22) carrier	France	1
1992	Simi et al. The unbalanced offspring of the male carriers of the 11q;22q translocation: nondisjunction at meiosis II in a balanced spermatocyte. Abstract: https://link.springer.com/article/10.1007/BF00215688	1-female age 10 Cleft palate, Micrognathia, Microcephaly Craniofacial asymmetry, Low set ears with preauricular tags, Strabismus, Iris coloboma Ectopic anus Father t(11;22) carrier	Italy	1
1991	Dean et al. Apparent Fryns' syndrome and aneuploidy: evidence for a disturbance of the midline developmental field. Abstract: https://onlinelibrary.wiley.com/doi/abs/10.1111/j.1399-0004.1991.tb03108.x	1-boy Left diaphragmatic hernia, Rocker bottom feet Rectal atresia,Cryptorchidism, Micropenis Hypoplastic lungs, Dandy-Walker malformation Mother t(11;22) carrier	UK	1

1991	Clark. A cytogenetic abnormality in tuberous sclerosis: Report of an affected infant with 47, XX, +der22,t(11;22)(q23.3;q11.2)mat. Abstract: https://nyaspubs.onlinelibrary.wiley.com/doi/10.1111/j.1749-6632.1991.tb37765.x	1-girl Cleft palate, Dysplastic ear, Micrognathia Preauricular tags, Excess nuchal skin Hypoplasia of spleen and kidneys Cardiac rhabdomyomas Olfactory nerves and sulci absent Brainstem and cerebellar structures hypoplastic Tuberous sclerosis Mother t(11;22) carrier	USA	1
1990	Katafuchi et al. Partial trisomy 22 with Dandy-Walker malformation. Abstract: https://onlinelibrary.wiley.com/doi/abs/10.1111/j.1442-200X.1990.tb00883.x	1-boy Prenatal diagnosis of Dandy-Walker malformation, Aplasia of right kidney Severe intrauterine growth retardation Hypotonia, Psychomotor delay Low set and malformed ears with preauricular pits, Micrognathia, Excess neck skin Long digits, Contracture of the right ankle joint Micropenis, Atrial septal defect, ventricular septal defect, marked pulmonary hypertension MRI showed cystic dilatation of the fourth ventricle, hypoplasia of the cerebellum with agenesis of the cerebellar vermis and upald extension of the third ventricle (indicating DWM)	Japan	1
1990	Abeliovich & Carmi. The translocation 11q;22q; A novel unbalanced karyotype. https://onlinelibrary.wiley.com/doi/abs/10.1002/ajmg.1320370227https://onlinelibrary.wiley.com/doi/abs/10.1002/ajmg.1320370227	1-boy Pregnancy terminated, Malformed ears Micrognathia, Dilatation of the cerebral ventricles	Israel	1
1989	Karimi-Nevada et al..	1-female	Iran	1

	Reconsideration of the cat eye syndrome: Reciprocal translocation t(11;22) leading to partial trisomy of 11q and 22. Full text: http://mjiri.iums.ac.ir/files/site1/user_files_e9487e/azimi-A-10-298-802-c9643e0.pdf	Cleft palate, Preauricular fistula Atrial septal defect, Ventricular septal defect Pulmonary stenosis, Right ventricular hypertrophy, Passed at 23 months due to respiratory infection		
1989	Lockwood et al. Not all chromosome imbalance resulting from the 11q;22q translocation is due to 3:1 segregation in first meiosis. Abstract: https://link.springer.com/article/10.1007%2FBF00285174	1-boy Father t(11;22) carrier Child had karyotype: 47,XY,t(11;22)(q23.3;q11.2),+der(22)t(11;22)(q23.3;q11.2)pat Clinical picture consistent with other cases	USA	1
1987	Sou et al. Prenatal diagnosis of partial trisomy 22 derived from a maternal t(11;22)(q23;q11), Full text: https://www.jstage.jst.go.jp/article/tjem1920/153/4/153_4_389/_pdf	1 case - fetal demise 23 weeks Mother 11/22 carrier Microcephaly, Cleft palate Malrotation of sigmoid and transverse colon	Japan	1
1987	Pattemore et al. The neonatal recognition of partial 11q trisomy (previously ‘Trisomy 22’). Abstract: https://onlinelibrary.wiley.com/doi/abs/10.1111/j.1440-1754.1987.tb00245.x	1-boy Micrognathia and receding tongue Cleft palate, Hypotonic, Absent left kidney Vesicoureteral reflux with intrarenal reflux Severe bilateral hearing loss, Ventricular septal defect, Dislocated hips, Growth at age 3 below 3rd percentile, Myopia, Abnormally shaped teeth, Severe developmental delay Mother t(11;22) carrier	New Zealand	1
1987	Noir et al. [11q trisomy: apropos of 2 cases] [Article in French]. https://pubmed.ncbi.nlm.nih.gov/3438103/	1-boy - age 5 Ambiguous sexuality Micrognathia hypotonia	France	1

1986	Beedgen et al. Partial trisomy 22 and 11 due to a paternal 11;22 translocation associated with Hirschsprung disease. Abstract: https://link.springer.com/article/10.1007/BF00446075	1-boy Hypotonia, Heart murmur, Severe micrognathia Excess neck skin, Micropenis, Bilateral cryptorchidism, Anal stenosis, Bilateral cervical ribs, Atrial septal defect, Agenesis of right kidney, Right sided hydronephrosis and hydroureter, Chronic renal insufficiency (solitary kidney) with chronic metabolic acidosis and anaemia, Atrial septal defect, Hirschsprung disease, Recurrent urinary tract infections Psychomotor delay Father t(11;22) carrier	Germany	1
1986	Tovo et al. Thymic hormone dependent immunodeficiency in an infant with partial trisomy of chromosome 22. Abstract: https://europepmc.org/article/med/3798528	1-boy Strabismus, Micrognathia with receding chin High arched palate, Bifid uvula, Low set and malformed ears, Bilateral preauricular tags Bilateral hip dislocation, Feeding and swallowing difficulties, Recurrent gastroenteritis and upper respiratory tract infections Optic nerve atrophy, Left renal aplasia Mother t(11;22) carrier Primary combined immunodeficiency disease (CID)	Italy	1
1986	Lin et al. Congenital heart disease in supernumerary der(22),t(11;22) syndrome. Abstract: https://onlinelibrary.wiley.com/doi/abs/10.1111/j.1399-0004.1986.tb01254.x	Review of 85 published cases		
1983	Iselius et al. The 11q;22q translocation: A collaborative study of	1 - boy - newborn - Italy Micrognathia, bilateral preauricular pits and	International	12

	20 new cases and analysis of 110 families. Abstract: https://link.springer.com/article/10.1007/BF00292366	tags, small penis, imperforate anus, massive left-sided diaphragmatic hernia, complete endocardial cushion defect, left ureteral stenosis, absence of the olfactory bulbs and tracts and cerebellar vermis 2 - girl, Germany Microcephaly, microretrognathia, low set and malformed ears, ear tags, dysplastic nails, hyperflexible joints, left-sided inguinal hernia, hypoplastic labia majora and minora, hypotonia, repeated ear and respiratory infections, 3 - girl -3 ½ years - Germany Micrognathia, ear tags, cleft palate, hypertonia, anal atresia, strabisumus, anomalies of the teeth, dislocated hips, seizures 4 - boy - 20 months - France Severely delayed physical development, hypotonia, ventricular septal defect, cleft palate, micrognathia, ear tag, left-sided inguinal hernia 5 - boy - 4 years - Japan Microcephaly, cleft palate, micrognathia, right inguinal hernia, micropenis, ventricular septal defect, dislocated hips, hypotonia, seizures, recurrent pneumonia 6 - sex unidentified - 10 months - Canada Bilateral ear pits, cleft palate, microcephaly, atrial septal palate, mild aortic valve stenosis, moderate pulmonary valve stenosis, bilateral dislocation of hips, severe psychomotor delay		

		7 - boy, 3 ½ years USA Growth below 3rd percentile, microcephaly, severe psychomotor delay, hypotonia, right facial palsy, skin tag and pit on left ear, malformed right ear with several pits, high arched palate, atrial septal defect 8 - sex unidentified - age 6 - Italy Microcephaly, delayed development, hypotonia, torticollis, trigonocephaly, scoliosis, dysplastic teeth 9 - girl -age 4 - Finland Cleft palate, ventricular septal defect, growth below 3rd percentile, severely delayed physical and mental development 10 - boy - age 15 - USA Weight below 3rd percentile, bilateral ear pits, atrial septal defect, unilateral renal agenesis, bilateral cryptorchidism, hypospadias, small penis, profound mental delay 11 - boy - 16 months - Sweden Hypotonia, cleft palate, extension defect in elbows, kyphosis, anal stenosis, atrial and ventricular septal defects, left sided superior vena cava, delayed boy and psychomotor development, hearing defect, renal insufficiency, delayed dentition. 12 - boy - 18 months - Sweden Dysplastic ears with tags, atresia of the right		

		external auditory canal, left cheek fibroma, cleft palate, bilateral dislocation of hips, contractures of knees and elbows, left sided pes equino-varus, slight hypospadias, delayed psychomotor development		
1981	Schinzel et al. Incomplete trisomy 22. Familiar 11/22 translocation with 3:1 meiotic disjunction. Delineation of a common clinical picture and report of nine new cases from six families. Abstract: https://link.springer.com/article/10.1007%2FBF00274675	1-boy - 15 years Anal stenosis, bilateral hip luxation, growth below 3rd percentile, trigonocephaly and asymmetry of the skull, left esotropia, irregular teeth, submucous cleft palate, bifid uvula, short mandible, bilateral preauricular skin tags, thoracic kyphosis, absence of left kidney, delayed bone age, 2-boy, age 14 Absent left auricle, cryptorchidism, anal atresia with fistula, severely delayed. Growth below 3rd percentile, malaligned teeth with enamel hypoplasia, submucous cleft palate, bifid uvula, preauricular tag, skin appendance replacing left auricle, no left ear canal, heart murmur, hypotonia, micropenis, hypoplastic scrotum, anterior rotation of left kidney, mild aortic stenosis, sclerosis of the skill, bone age delayed. 3-girl, age 7 Preauricular skin tag, bilateral hip subluxation, recurrent upper respiratory tract infections and otitis, growth below 3rd percentile, delayed dentition, high and narrow palate, poorly	Germany and Switzerland	9

		shaped right ear, excess neck skin, hypotonia 4-boy - age 4 Small penis, hypoplastic scrotum with no palpable testes, microcephaly, high narrow palate, preauricular fistula, scoliosis, left clubfoot, myopia, astigmatism, esotropia, severe conductive hearing loss, recurrent respiratory and ear infections, severe delay, scoliosis, growth below 3rd percentile, brachycephaly, left facial palsy, severe delay in bone growth, right kidney smaller than normal, psychomotor delay. 5-female newborn Left preauricular tag, short mandible, cleft palate, anteriorly placed anus, hypotonia, congenital dysplasia of the left hemidiaphragm,hypoplasia of the left lung and bronchial tree, heart defects, bilateral renal hypoplasia, unusual lobulation of the liver, duplication of the uterus and vagina. 6-girl -newborn Cleft palate, poorly formed auricles, redundant neck skin, flexion contractures of knees and ankles, 13 thoracic vertebrae and ribs, malrotation of the small bowel, hypotonia, hypoplasia of the corpus callosum and other brain abnormalities, cardiac abnormalities, hypoplasia of the left lung, malrotation of small bowel, one supernumerary spleen, hypoplasia of both kidneys.		

		7-boy newborn Receding mandible, cleft palate, seizures 8-boy, 3 months Small mouth and tongue, cleft palate, receding mandible, bilateral preauricular pits and tags, narrow external ear canals, bilateral inguinal hernias, micropenis, glandular hypospadias, undescended testes, limited abduction in the left hip, hypotonia, joints could be over-extended, hydronephrosis and hydroureter of the kidney and bilateral vesicoureteral reflux, agenesis of the intrahepatic bile duct, biliary cirrhosis. 9-boy, newborn Hypotonic, small mandible, cleft palate, poorly formed ears.		
1981	Anneren & Gustavsen. Trisomy 22 syndrome in a 26-year old female-A follow-up examination. Full text: https://onlinelibrary.wiley.com/doi/epdf/10.1111/j.1601-5223.1981.tb01733.x	1-girl age 26 Cleft palate, Micrognathia, Subluxation of the right hip, Microcephaly, Strabismus, Malformed ears, Hearing impairment, Microcephaly 2-girl - age 6 Cleft palate, Micrognathia, Malformed ears Hearing impairment, Facial asymmetry Rectal stenosis, Hypotonia, Microcephaly Strabismus,Atrial septal defect	Sweden	2
1980	Najafzadeh & Dumars. Duplication of distal 11q and 22p occurrence in two unrelated families. Abstract: https://onlinelibrary.wiley.com/doi/epd	1 - girl - age 2 Micrognathia, preauricular tags, cleft palate, tube fed due to swallowing issues, repeated aspiration, pneumonia and convulsions,	USA	2

	f/10.1002/ajmg.1320080313	hypotonia delayed development. Mother t(11;22) carrier 2 - male - Lumbosacral meningomyelocele, bilateral club feet, single umbilical artery, preauricular tags and pits, micrognathia, submucous cleft palate, ventricular septal defect, small penis, bilateral indirect hernia, congenital hip dislocation, hypotonia, 11 ribs on the left, absent midportion of the right clavicle, bilateral sensorineural hearing loss. Mother t(11;22) carrier		
1980	Pangalos et al. [Partial 11q trisomy due to missegregation of maternal t(11;22) (q23;q11.1) translocation (author's transl)] [Article in French] Abstract: https://pubmed.ncbi.nlm.nih.gov/7443445/	1-girl 4 months Micrognathia, Cleft palate, Ear abnormalities Heart defects Mother t(11;22) carrier	Greece	1
1980	Chauveau et al. [A new observation of trisomy 11q due to t(11,22)(q23.1; q11.1) mat (author's transl)] [Article in French]. Abstract: https://pubmed.ncbi.nlm.nih.gov/6971598/	1-male Hypotonia, micrognathia, heart defect, hypoplastic penis	France	1
1980	Fraccaro et al. The 11q;22q translocation: A European collaborative analysis of 43 cases. Abstract: https://link.springer.com/article/10.10	20 boys, 12 girls Age range newborn to age 20 Common findings listed: low birth weight, delayed development, hypotonia, microcephaly, craniofacial asymmetry,	European collaborative	32 unbalanced cases

	07/BF00281567	malformed ears with pits and tags, cleft palate, micro/retrognathia, strabismus, congenital heart disease, cryptorchism, congenital dislocation of the hip.		
1980	Biederman et al. Tertiary trisomy (22q11q),47,+der(22),t(11;22). Abstract: https://link.springer.com/article/10.1007/BF00273491	1-girl - age 12 Cleft palate Anteriorly displaced anus, Valgus deformity of the lower extremities, Seizures Congenital heart disease/atrial septal defect Growth below 3rd centile, Kyphoscoliosis Hypoplastic external genitalia, Left hemiplegia with marked spasticity on left Mother t(11;22) carrier	Canada	1
1979	Nakai, et al. Partial trisomy of 11 and 22 due to familial translocation t(11;22)(q23q11) inherited in three generations. Abstract: https://link.springer.com/article/10.1007/BF00283408	1-girl Cleft palate, Heart disease, Congenital dislocation of hip, Mother and maternal grandfather both carriers	Japan	1
1979	Narahara et al. A case of partial trisomy 22 resulting from maternal 11/22 translocation. Full text: https://www.nature.com/articles/jhg197927.pdf	1-girl -newborn Microcephaly, Right preauricular skin tag and pits, Micrognathia, Cleft palate, Heart murmur Limited abduction of hip joints, Delayed development, Growth below 3rd percentile Atrial septal defect.Passed away at 8 months. Mother t(11;22) carrier	Japan	1
1979	Pai et al. Syndromes due to chromosomal abnormalities: Partial trisomy 22, interstitial deletion of the long arm of 13, and trisomy 8. No abstract available.	1-girl Cleft palate, Dysplastic left hip, Deformity of right foot, Severe hypotonia, Recurrent respiratory and ear infections, Severe developmental delay, Persistent growth delay	USA	1

	https://pubmed.ncbi.nlm.nih.gov/491337/	Seizures Mother t(11;22) carrier		
1979	Fryns et al. Partial trisomy 22q with elevated arylsulfatase-a activity. Abstract: https://europepmc.org/article/med/43111	1-male - age 2 Hypotonia, Right sided inguinal hernia, Left facial palsy, Small penis, hypoplastic scrotum and small testes, Frequent upper respiratory infections, Tooth development delayed Strabismus	Belgium	1
1978	Iselius & Faxelius. Trisomy 22 in a newborn girl with multiple malformations. Full text: https://onlinelibrary.wiley.com/doi/epdf/10.1111/j.1601-5223.1978.tb01286.x	Micrognathia, Anal atresia, Persistent left superior vena cava, Ventricular septal defect Patent ductus arteriosus, Recto-vaginal fistula Small kidneys	Sweden	1
1978	Feldman & Sparkes. The problem of partial trisomy 22 reconsidered. Abstract: https://link.springer.com/article/10.1007/BF00277580	1 - girl - 3 years Abnormal appearance, Heart murmur Dislocated hips, Hypotonia, Poor feeding Delayed development and poor motor skills Coxa valgus, Coarctation of the aorta Microcephaly, Micrognathia Mother t(11;22) carrier	USA	1
1977	Kessel & Pfeiffer. 47,XY,+der(22(11;22)(q23q12) Following balanced translocation t(11;22)(q23;q12)mat. Remarks on the problem of trisomy 22. Abstract https://link.springer.com/article/10.1007/BF00293781	1-boy age 10 Feeding issues, Failure to thrive, Unilateral hip dysplasia, Febrile seizures, Recurrent infections of ears and upper respiratory tract Cleft of the soft palate, Hyperextensible fingers and toes, Cryptorchidism, Micropenis, Deafness, Generalized muscle weakness Mother t(11;22)carrier	Germany	1
1977	Bowfinger, Shirley & Soukup. Cat	1-girl	USA	1

	Eye Syndrome: Partial trisomy 22 due to translocation in the mother. Abstract: https://jamanetwork.com/journals/jamapediatrics/article-abstract/507631	Preauricular skin tags, High palate, Excess neck skin, Heart murmur, pulmonary stenosis Single kidney, Imperforate anus, Limited hip abduction, Mother balanced t(11;22) carrier		
1976	Fu et al. Structural aberrations of the long arm of chromosome no. 22: Report of a family with translocation t(11;22)(q25q11). Abstract: https://onlinelibrary.wiley.com/doi/abs/10.1111/j.1399-0004.1976.tb00057.xdf	1-boy Low-set ears Pulmonary stenosis, double aortic arch and atrial septal defect	USA	1
1976	Oakley et al. Balanced translocation, impaired sperm motility, and offspring anomaly. Full text: https://adc.bmj.com/content/archdischild/51/8/638.full.p	1- Cleft palate, Enlarged liver and spleen Excess neck skin/swelling, Ventricular septal defect, Hypotonia Father t(11;22) carrier	UK	1
1976	Ayraud et al. [Trisomy 11q (q23.1 - qter) through maternal translocation t(11;22) (q23.1;q11.1). A new case] [Article in French]. Abstract: https://pubmed.ncbi.nlm.nih.gov/1084126/	1-boy age 4 Hypotonia, Preauricular fistula, Micrognathia cleft palate,	France	1
1976	Noel et al. [Partial trisomy of the long arm of the chromosome 11 by malsegregation of a maternal translocation t(11;22)(q23 1;q1 11)] [Article in French]. Abstract: https://pubmed.ncbi.nlm.nih.gov/1085603/	1-male 10 months Diaphragmatic hernia Seizures Mother t(11;22) carrier	France	1

1976	Emanuel et al. Abnormal chromosome 22 and recurrence of trisomy 22-syndrome. Full text: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC1013477/	1-girl newborn Micrognathia, High arched palate Ear abnormalities, Cardiac abnormalities Brain abnormalities 2- boy age 5 Cleft palate, Ear abnormalities, Dislocated left hip, Small penis, Imperforate anus	USA	2
1975	Giraud et al. [Partial trisomy 11q and familial translocation 11--22 (author's transl)] [Article in French]. https://pubmed.ncbi.nlm.nih.gov/1176125/	Siblings	France	1
1975	Aurias, et al.[Deux cas de trisomie 11q(q231->qter) par translocation t(11;22)(q231;q111) dans deux familles différentes.] [Article in French]. Abstract: https://pubmed.ncbi.nlm.nih.gov/1080981/	1-boy Micrognathia, cleft palate, Pierre Robin sequence, malformed ears, cardiac issues, mother t(11;22) carrier 1-girl Cleft palate, micrognathia, ear malformations, aortic stenosis, cardiac issues, mother t(11;22) carrier	France	2
1975	Laurent et al. [2 cases of trisomy 11q(q23.2-- qter) with the same abnormality of external genitalia] [Article in French] Abstract: https://pubmed.ncbi.nlm.nih.gov/1080980/	1- boy Micropenis Cleft palate, micrognathia, ear abnormalities, heart defect		