CURRICULUM VITAE
Dott.ssa MARIA CRISTINA DIGILIO

Data di nascita: 14 maggio 1961
Stato civile: nubile
Indirizzo permanente: Viale Marco Polo 77, 00154 Roma. Tel. 06/5743229
Indirizzo ospedale: Ospedale Pediatrico Bambino Gesù, U.O. Genetica Medica, Piazza S.Onofrio 4, 00165, Roma. Tel. 06/68592227

TITOLI DI STUDIO:

- Laurea in Medicina e Chirurgia, presso l'Università degli Studi di Roma La Sapienza, 24/10/86
- Abilitazione all'esercizio della professione medico-chirurgica, presso l'Università degli Studi La Sapienza di Roma, Novembre 86
- Specializzazione in Pediatria, presso l'Università degli Studi La Sapienza di Roma, 27/6/90
- Specializzazione in Genetica Medica, presso l'Università Cattolica del Sacro Cuore di Roma, 9/6/95
- Iscrizione all'Ordine dei Medici Chirurghi di Roma, dal 2/4/87 a tutt'oggi

ATTIVITA' PROFESSIONALE

- Servizio in qualità di Dirigente Medico di I livello presso l'U.O. di Genetica Medica dell'Ospedale Bambino Gesù di Roma: dal 5/8/94 a tutt'oggi
- Responsabile dell'Alta Specializzazione in Dismorfologia presso lo stesso ospedale: dall' 1/1/2002 a tutt'oggi

STAGES DI FORMAZIONE ALL'ESTERO

- Stage presso il Baltimore-Washington Infant Study Group, University of Maryland, Baltimore, USA, novembre 2002

ATTIVITA' DIDATTICA

- Corso Universitario per Infermieri, Università di Tor Vergata, Roma, Anni accademici: 2000-2001 a tutt'oggi
- Corsi di Scuola Medica Ospedaliera "Sindromi genetiche in Pediatria: Clinica e Laboratorio", Anni accademici: 2002-2003, 2005-2006.

ISCRIZIONE A SOCIETA' MEDICHE

- Ordine dei Medici di Roma e Provincia
- Società Italiana di Genetica Umana (SIGU)
- Società Italiana di Pediatria (SIP)
- SIMGePeD
- Società Italiana di Endocrinologia Pediatrica (SIEDP)

REVISORE DELLE SEGUENTI RIVISTE

- American Journal of Medical Genetics
- Clinical Genetics
- American Journal of Cardiology
- European Journal of Echocardiography

LETTURE / CONFERENZE TENUTE ALL'ESTERO SU INVITO

- International Meeting on the etiology and morphogenesis of congenital heart diseases, Tokyo, Japan, November 7-9, 2002.
- 12th International Scientific Congress on the DiGeorge syndrome, Strasbourg, France, July 7-9, 2006
- The fifth International 22q11.2 deletion syndrome Conference, Marseille, France, July 10-11, 2006
- Rare disorders of the MAPK pathway: Current status / future directions. Barcelona, Spain, May 30-31, 2008

RELATORE SU INVITO in circa 25 meetings e congressi italiani

ARTICOLI IN GIORNALI INDEX

2008

Digilio MC, Calzolari F, Capolino R, Toscano A, Sarkozy A, de Zorzi A, Dallapiccola B, Marino B. Congenital heart defects in patients with oculo-auriculo-vertebral spectrum (Goldenhar syndrome). Am J Med Genet 2008;146A:1815-1819.

Carotti A, **Digilio MC,** Piacentini G, Saffirio C, Di Donato RM, Marino B. Cardiac defects and results of cardiac surgery in 22q11.2 deletion syndrome. Dev Disabil Res Rev 2008;14:35-42.

Digilio MC, Sarkozy A, Capolino R, Chiarini Testa MB, Esposito G, de Zorzi A, Cutrera R, Marino B, Dallapiccola B. Costello sindrome: clinical diagnosis in the first year of life. Eur J Pediatr 2008;167:621-628.

Sarkozy A, **Digilio MC**, Dallapiccola B. LEOPARD syndrome. Orphanet J Rare Dis 2008; 3: 13.

Digilio MC, Marino B, Dallapiccola B. Deletion 22q11 and isolated congenital heart disease. Int J Cardiol 2008; 123:364-365.

Limongelli G, Sarkozy A, Pacileo G, Calabrò P, **Digilio MC,** Maddaloni V, Gagliardi G, Di Salvo G, Iacomino M, Marino B, Dallapiccola B, Calabrò R. Genotype-phenotype analysis and natural history of left ventricular hypertrophy in LEOPARD syndrome. Am J Med Genet 2008;146A:620-628.

Digilio MC, Capolino R, Dallapiccola B. Autosomal dominant transmission of nonsyndromic diastasis recti and weakness of the linea alba. Am J Med Genet 2008;146A:254-256.

Casaccia G, **Digilio MC**, Seymandi PL, Bagolan P. Congenital diaphragmatic hernia in CHRGE syndrome. Pediatr Surg Int 2008;24:375-378.

Limongelli G, Pacileo G, Melis D, Calabrò P, **Digilio MC**, Sarkozy A, Maddaloni V, Capozzi G, Sebastio G, Andria G, Calabrò R. Trisomy 18 and hypertrophic cardiomyopathy in an 18-year-old woman. Am J Med Genet 2008;146A:327-329.

2007

Pandit B, Sarkozy A, Pennacchio LA, Carta C, Oishi K, Martinelli S, Pogna EA, Schackwitz W, Ustaszewska A, Landstrom A, Bos JM, Ommen SR, Esposito G, Lepri F, Faul C, Mundel P, Lopez Siguero JP, Tenconi R, Selicorni A, Rossi C, Mazzanti L, Torrente I, Marino B, **Digilio MC**, Zampino G, Ackerman MJ, Dallapiccola B, Tartaglia M, Gelb BD. Gain-of-function RAF1 mutations cause Noonan syndrome and LEOPARD syndromes with hypertrophic cardiomyopathy. Nat Genet 2007;8:1007-1012.

Tartaglia M, Pennacchio LA, Zhao C, Yadav KK, Fodde V, Sarkozy A, Pandit B, Oishi K, Martinelli S, Schackwitz W, Ustaszewska A, Martin J, Bristol J, Carta C, Lepri F, Neri C, Vasta I, Gibson K, Curry CJ, Siguero JPL, **Digilio MC**, Zampino G, Dallapiccola B, Bar-Sagi D, Gelb BD. Gain-of-function SOS1 mutations cause a distintive form of Noonan syndrome. Nat Genet 2007;39:75-79.

Limongelli G, Pacileo G, Marino B, **Digilio MC**, Sarkozy A, Elliott P, Versacci P, Calabrò P, de Zorzi A, Di Salvo G, Syrris P, Patton M, McKenna WJ, Dallapiccola B, Calabrò R. Prevalence and clinical significance of cardiovascular abnormalities in patients with LEOPARD syndrome. Am J Cardiol 2007;100:736-741.

Piacentini G, Marino B, **Digilio MC**. Familial recurrence risk of discrete memebranous subaortic stenosis. J Thorac Cardiovasc Surg 2007;134:818-819.

Limongelli G, Pacileo G, **Digilio MC**, Calabrò P, Di Salvo G, Rea A, Miele T, Frigiola A, Sarkozy A, Dallapiccola B, Marino B, Calabrò R. Severe, obstructive biventricular hypertrophy in a patient with Costello sindrome: Clinical impact and management. Int J Cardiol 2007;Epub a head of print.

Calcagni G, **Digilio MC**, Sarkozy A, Dallapiccola B, Marino B. Familial recurrence of congenital heart disease: An overview and review of the literature. Eur J Pediatr 2007;166:111-116.

Saffirio C, Marino B, **Digilio MC**. GATA4 as candidate gene for pericardial defects. Ann Thorac Surg 2007;84:2137.

Sarkozy A, Schirinzi A, Lepri F, Bottillo I, De Luca A, Pizzuti A, Tartaglia M, **Digilio MC**, Dallapiccola B. Clinical lumping and molecular splitting of LEOPARD and NF1/NF1-Noonan sindromes. Am J Med Genet 2007;143A:1009-1011.

Piacentini G, **Digilio MC**, Sarkozy A, Placidi S, Dallapiccola B, Marino B. Genetics of congenital heart diseases in syndromic and non-syndromic patients: new advances and clinical implications. J Cardiovasc Med 2007;8:7-11.

Cambiaso P, Orazi C, **Digilio MC**, Loche S, Capolino R, Tozzi A, Fredda P, Cappa M. Thyroid morphology and subclical hypothyroidism in children and adolescents with Williams sindrome. J Pediatr 2007;150:62-65.

2006

Digilio MC, Sarkozy A, de Zorzi A, Pacileo G, Limongelli G, Mingarelli R, Calabrò R, Marino B, Dallapiccola B. LEOPARD syndrome: clinical diagnosis in the first year of life. Am J Med Genet A 2006;140: 740-746.

Digilio MC, Sarkozy A, Pacileo G, Limongelli G, Marino B, Dallapiccola B. PTPN11 gene mutations: linking the Gln510Glu mutation to the "LEOPARD syndrome phenotype". Eur J Pediatr 2006;165:803-5.

Digilio MC, Dallapiccola B, Marino B. Atrioventricular canal defect in Bardet-Biedl syndrome: Clinical evidence supportino the link betweeen atrioventricular canal defect and polydactyly syndromes with ciliary dysfunction. Genet Med 2006;8:536-538.

Carta C, Pantaleoni F, Bocchinfuso G, Stella L, Vasta I, Sarkozy A, **Digilio MC**, Palleschi A, Pizzuti A, Grammatico P, Zampino G, Dallapiccola B, Gelb BD, Tartaglia M. Germline missense mutations affecting KRAS Isoform B are associated with severe Noonan syndrome phenotype. Am J Hum Genet 2006 Jul; 79(1): 129-35.

Howald C, Merla G, **Digilio MC**, Amenta S, Lyle R, Deutsch S, Choudhury U, Bottani A, Antonarakis SE, Fryssira H, Dallapiccola B, Reymond A. Two high throuput technologies to detect segmental aneuploidies identify new Williams-Beuren syndrome patients with atypical deletions. J Med Genet 2006;43:266-273.

Sarkozy A, Lepri F, Marino B, Pizzuti A, **Digilio MC**, Dallapiccola B. Additional evidence that PTPN11 mutations play only a minor role in the pathogenesis of non-syndromic atrioventricular canal defect. Am J Med Genet 2006;140:1970-1972.

Vergara P, **Digilio MC**, De Zorzi A, Di Carlo D, Capolino R, Rimini A, Pelegrini M, Calabrò R, Marino B. Genetic heterogeneity and phenotypic anomalies in children with atrioventricular canal defect and tetralogy of Fallot. Clin Dysmorphol 2006;15:65-70.

Calcagni G, **Digilio MC**, **Capolino R**, Dallapiccola B, Marino B. Concordant familial segregation of atrial septal defect and Axenfeld-Rieger anomaly in father and son. Clin Dysmorphol 2006;15: 203-6.

Michielon G, Marino B, Formigari R, Gargiulo G, Picchio F, **Digilio MC**, Anaclerio S, Oricchio G, Sanders SP, Di Donato RM. Genetic syndromes and outcome after surgical correction of tetralogy of Fallot. Ann Thorac Surg 2006;81:968-75.

Restivo A, Sarkozy A, **Digilio MC**, Dallapiccola B, Marino B. 22q11 deletion syndrome: a review of some developmental biology aspects of the cardiovascular system. J Cardiovasc Med (Hagerstown) 2006;7:77-85.

Sarkozy A, **Digilio MC,** Marino B, Mingarelli R, Tartaglia M, Dallapiccola B. Noonan's syndrome and related disorders: clinical-molecular update and guidelines. Ital J Pediatr 2006 Oct; 32(5):145-155.

Placidi S, **Digilio MC**, Marino B. Types of cardiac defects in children with Down's syndrome. Cardiol Young 2006;16:198-199.

Vergara P, **Digilio MC**, Limongelli G, Carotti A, Toscano A, Santoro G, Calabrò R, Marino B. Familial recurrence of anomalous origin of the right pulmonary artery from the aorta. Am J Med Genet 2006;140:794-796.

2005

Digilio MC, Marino B, Capolino R, Angioni A, Sarkozy A, Roberti MC, Conti E, de Zorzi A, Dallapiccola B. Familial recurrence of nonsyndromic congenital heart defects in

first degree relatives of patients with deletion 22q11.2. Am J Med Genet 2005;134:158-164.

De Luca A, Bottillo I, Sarkozy A, Carta C, Neri C, Bellacchio E, Schirinzi A, Conti E, Zampino G, Battaglia A, Majore S, Rinaldi MM, Carella M, Marino B, Pizzuti A, **Digilio MC**, Tartaglia M, Dalla piccola B. Am J Hum Genet 2005;77:1092-1101.

Versacci P, **Digilio MC**, Sauer U, Dallapiccola B, Marino B. Absent pulmonary valve with intact ventricular septum and patent ductus arteriosus: a specific cardiac phenotype associated with deletion 18q syndrome. Am J Med Genet 2005;138A:185-186.

Digilio MC, Dallapiccola B, Marino B. Association of deletion 22 and trisomy 21: a likely random association in patients with conotruncal defects. Am J Med Genet 2005;134A:1-2.

Digilio MC, Capolino R, Marino B, Sarkozy A, Dallapiccola B. Congenital intrahepatic portosystemic venous shunt: an unusual feature in LEOPARD sindrome and in neurofibromatosis type 1. Am J Med Genet 2005;134A: 457-458.

Sarkozy A, Conti E, Neri C, D'Agostino R, **Digilio MC**, Esposito G, Toscano A, Marino B, Pizzuti A, Dallapiccola B. Spectrum of atrial septal defects associated with mutations of NKX2.5 and GATA4 transcription factors. J Med Genet 2005;42:e16.

Piacentini G, **Digilio MC**, Capolino R, De Zorzi A, Toscano A, Sarkozy A, D'Agostino R, Marasini M, Russo MG, Dallapicccola B, Marino B. Familial recurrence of heart defects in subjects with congenitally corrected transposition of the great arteries. Am J Med Genet A 2005;137:176-180.

Sarkozy A, Esposito G, Conti E, **Digilio MC**, Marino B, Calabrò R, Pizzuti A, Dallapiccola B. CRELD1 and GATA4 gene analysis in patients with nonsyndromic atrioventricular canal defects. Am J Med Genet 2005;139:236-238.

Mingarelli R, Zuccarello D, **Digilio MC**, Dallapiccola B. A new observation of acro-cardio-facial syndrome substantiates interindividual clinical variability. Am J Med Genet 2005; 136:84-86.

Novelli A, Ceccarini C, Bernardini L, Zuccarello D, **Digilio MC**, Mingarelli R, Dallapiccola B. Pure trisomy 19p sindrome in an infant with an extra ring chromosome. Cytogenet Menome Res 2005;111:182-185.

Sarkozy A, Conti E, D'Agostino R, **Digilio MC**, Formigari R, Picchio F, Marino B, Pizzuti A, Dallapiccola B. ZFPM2/FOG2 and HEY2 genes analysis in nonsyndromic tricuspid atresia. Am J Med Genet 2005;133A:68-70.

2004

Brancati F, D'Avanzo MG, **Digilio MC**, Sarkozy A, Biondi M, De Brasi D, Mingarelli R, Dallapiccola B. KBG syndrome in a cohort of Italian patients. Am J Med Genet A 2004;131:144-149.

Novelli A, Ceccarini C, Bernardini L, Zuccarello D, Caputo V, **Digilio MC**, Mingarelli R, Dallapiccola B. High frequency of subtelomeric rearrangements in a cohort of 92 patients with severe mental retardation and dysmorphism. Clin Genet 2004;66:30-38.

Sarkozy A, Conti E, **Digilio MC**, Marino B, Morini E, Pacileo G, Wilson M, Calabrò R, Pizzuti A, Dallapiccola B. Clinical and molecular analysis of 30 patients with multiple lentigines LEOPARD syndrome. J Med Genet 2004;41:e68.

Digilio MC, Pacileo G, Sarkozy A, Limongelli G, Conti E, Cerrato F, Marino B, Pizzuti A, Calabrò R, Dallapiccola B. Familial aggregation of genetically heterogeneous hypertrophic cardiomyopathy: a boy with LEOPARD syndrome due to PTPN11 mutation and his nonsyndromic father lacking PTPN11 mutations. Birth Def Res A Clin Mol Teratol 2004;70:95-98.

Digilio MC, Capolino R, Versacci P, Marino B. Polyvalvular heart disease associated with short stature, facial anomalies, and mental retardation: an additional familial report. Am J Med Genet 2004;127:101-103.

Digilio MC, Torrente I, Goodship JA, Marino B, Novelli G, Giannotti A, Dallapiccola B. Ellis-van Creveld syndrome with hydrometrocolpos is not linked to chromosome arm 4p or 2p. Am J Med Genet 2004;126:319-323.

Nobili V, Marcellini M, Devito R, Capolino R, Viola L, **Digilio MC**. Hepatic fibrosis in Kabuki syndrome. Am J Med Genet A 2004;124:209-212.

Anaclerio S, Di Ciompo V, Michielon G, **Digilio MC**, Formigari R, Picchio FM, Gargiulo G, Di Donato R, De Ioris MA, Marino B. Conotruncal heart defects: impact of genetic syndromes on immediate operative mortalità. Ital Heart J 2004;5:624-628.

2003

Digilio MC, Giannotti A, Dallapiccola B, Marino B. Postural anomaly of vthe head-neck-shoulder alignment in patients with deletion 22q11.2 (DiGeorge/velocardiofacial sindrome). Clin Genet 2003;64:447-448.

Pizzuti A, Sarkozy A, Newton AL, Conti E, Flex E, **Digilio MC**, Amati F, Gianni D, Tandoi C, Marino B, Crossley M, Dallapiccola B. Mutations of ZFPM2/FOG2 gene in sporadic cases of tetralogy of Fallot. Hum Mutation 2003;22:372-377.

Digilio MC, Angioni A, De Santis M, Lombardo A, Giannotti A, Dallapiccola B, Marino B. Specrum of clinical variability in familial deletion 22q11.2: from full manifestation to extremely mild clinical anomalies. Clin Genet 2003;63:308-313.

Sarkozy A, Conti E, Seripa D, **Digilio MC**, Grifone N, Tandoi C, Fazio VM, Di Ciommo V, Marino B, Pizzuti A, Dallapiccola B. Correlation between PTPN11 gene mutations and congenital heart defects in Noonan and LEOPARD syndromes. J Med Genet 2003;40:704-708.

Digilio MC, Giannotti A, Castro M, Colistro F, Ferretti F, Marino B, Dallapiccola B. Screening for celiac disease in patients with deletion 22q11.2 (DiGeorge/velo-cardio-facial syndrome). Am J Med Genet 2003;121:286-288

Digilio MC, Angioni A, Giannotti A, Dallapiccola B, Marino B. Truncus arteriosus and duplication 8q. Am J Med Genet 2003;121:79-81.

Digilio MC, Marino B, Giannotti A, Dallapiccola B, Opitz JM. Specific congenital heart defects in RSH/Smith-Lemli-Opitz syndrome: postulated involvement of the Sonic Hedghog pathway in syndromes with postaxial polydactyly or heterotaxia. Birth Defects Res A Clin Mol Teratol 2003;67:149-153.

Scattone A, Caruso G, Marzullo A, Piscitelli D, Gentile M, Bonadonna L, Balducci G, **Digilio MC**, Jenkner A, Camassei FD, Boldrini R, Nazzaro P, Pollice L, Serio G. Neoplastic disease and deletion 22q11.2: a multicentric study and report of two cases. Pediatr Pathol Mol Med 2003;22:323-341.

Marino B, Mileto F, **Digilio MC**. Tetralogy of Fallot with aortic valvular stenosis and deletion 22q11. Ann Thorac Surg 2003;75:2010-2011.

Tessa A, Salvi S, Casali C, Garavelli L, **Digilio MC**, Dotti MT, Di Ginadomenico S, Valoppi M, Grieco GS, Comanducci G, Bianchini , Fortini D, Federico A, Giannoti A, Santorelli FM. Six novel mutations of the RUNX2 gene in Italian patients with cleidocranial displasia. Hum Mut 2003;22:104.

Pastore A, Tozzi G, Gaeta LM, Giannotti A, Bertini E, Federici G, **Digilio MC**, Piemonte F. Glutathione metabolism and antioxidant enzymes in children with Down sindrome. J Pediatr 2003;142:583-585.

Cappa M, Bizzarri C, Colabianchi D, Ubertini G, **Digilio MC**, Cambiaso P. Growth and pubertal growth spurt in dysmorphic syndromes. J Pediatr Endocrinol Metab 2003;16 Suppl 2:317-320.

Conti E, Grifone N, Sarkozy A, Tandoi C, Marino B, **Digilio MC**, Mingarelli R, Pizzuti A, Dallapiccola B. DiGeorge subtypes of nonsyndromic conotruncal defects: evidence against a major role of TBX1 gene. Eur J Hum Genet 2003;11:349-351.

Pierdominici M, Mazzetta F, Caprini E, Marziali M, **Digilio MC**, Marino B, Aiuti A, Amati F, Russo G, Novelli G, Pandolci F, Luzi G, Giovannetti A. Biased T-cell receptor repertoires in patients with chromosome 22q11.2 deletion sindrome (DiGeorge sindrome/velocardiofacial sindrome). Clin Exp Immunol 2003;132:323.331.

Sarkozy A, Conti E, Esposito G, Pizzuti A, Dalla piccola B, Mingarelli R, Marino B, **Digilio MC**, Paoletti V. Nonsyndromic pulmonary valve stenosis ad the PTPN11 gene. Am J Med Genet 2003;116°:389-390.

2002

Digilio MC, Conti E, arkozy A, Mingarelli R, Dottorini T, Marino B, Pizzuti A, Dalla piccola B. Grouping the multiple-lentigines/LEOPARD and Noonan syndromes on the PTPN11 gene. Am J Hum Genet 2002;71:389-394.

Marino B, Capolino R, **Digilio MC**, Di Donato R. Transposition of the great ateries in asplenia and polysplenia phenotypes. Am J Med Genet 2002;110:292-294.

Patrono C, Dionisi-Vici C, Giannotti A, Bembi B, **Digilio MC**, Rizzo C, Purificato C, Martini C, Pierini R, Santorelli FM. Two novel mutations of the human delta7-sterol reductase (DHCR7) gene in children with Smith-Lemli-Opitz syndrme. Mol Cell Probes 2002;16:315-318.

Toscano A, Anaclerio S, **Digilio MC**, Giannotti A, Fariello G, Dallapiccola B, Marino B. Ventricular septal defect and deletion of chromosme 22q11:anatomical types and aortic arch anomalies. Eur J Pediatr 2002;161:116-117.

Marino B, **Digilio MC**, Versacci P, Anaclerio S, Dallapiccola B. Transposition of great arteries. Understanding its pathogenesis. Ital Heat J 2002;3 Suppl:154-160.

2001

Digilio MC, Casey B, Toscano A, Calabrò R, Pacileo G, Marasini M, banaudi E, Giannotti A, Dalla piccola B, Marino B. Complete transposition of the great arteries: Patterns of congenital heart disease in familial precurrence. Circulation 2001;104:2809-2814.

Digilio MC, Marino B, Toscano A, Giannotti A, Dallapiccola B. Congenital heart defects in Kabuki sindrome. Am J Med Genet 2001;100:269-274.

Digilio MC, Marino B, Cappa M, Cambiaso P, Giannotti A, Dallapiccola B. Auxological evaluation in patients with DiGeorge/velocardiofacial syndrome (deletion 22q11.2 syndrome). Genet Med 2001;3:30-33.

Digilio MC, Marino B. Genetic predisposition to ventricular septal defect in Down sindrome. Hum Genet 2001;109:463.

Marino B, **Digilio MC**, Toscano A, Anaclerio S, Giannotti A, Feltri C, De Ioris MA, Angioni A, Dallapiccola B. Anatomic patterns of conotruncal defects associated with deletion 22q11. Genet Med 2001;3:45-48.

Giordano U, Turchetta A, Giannotti A, **Digilio MC**, Virgilii F, Calzolai A. Exercise testing and 24-hour ambulatorial blood pressare monitoring in children with Williams syndrome. Pediatr Cardiol 2001;22:509-511.

Mangino M, Flex E, **Digilio MC**, Giannotti A, Dallapiccola B. Identification of a novel NOG gene mutation (P35S) in an Italian family with symphalangism. Hum Mut 2002;308.

Marino B, **Digilio MC**, Di Donato R. Health supervision for children with Down syndrome. Pediatrics 2001;108:1384.

Giannotti A, Tiberio G, Castro M, Virgilii F, Colistro F, Ferretti F, **Digilio MC**, Gambarara M, Dallapiccola B. Coeliac disease in Williams syndrome. J Med Genet 2001;38:767-768.

Anaclerio S, Marino B, Carotti A, **Digilio MC**, Toscano A, Gitto P, Giannotti A, Di Donato R, Dallapiccola B. Pulmonary atresia with ventricular septal defect: prevalence of deletion 22q11 in the different anatomic patterns. Ital Heart J 2001;2:384-387.

Gigante M, Matera MG, Seripa D, Izzo AM, Venanzi R, Giannotti A, **Digilio MC**, Gravina C, Lazzari M, Monteleone M, Dallapiccola B, Fazio VM. EXT-mutation analysis in Italian sporadic and hereditary osteochondromas. Int J Cancer 2001;95:378-383.

2000

Digilio MC, Marino B, Musolino AM, Giannotti A, Dallapiccola B. Familial recurrence of nonsyndromic interrupted aortic arch and truncus arteriosus with atrioventricular canal. Teratology 2000;61:329-331.

Digilio MC, Marino B, Giannotti A, Di Donato R, Dallapiccola B. Heterotaxy with left atrial isomerism in a patient with deletion 18p. Am J Med Genet 2000;94:198-200.

Giglio S, Graw SL, Gimelli G, Pirola B, Varone P, Voullaire L, Lerzo F, Rossi E, Della vecchia C, Bonaglia MC, **Digilio MC**, Giannotti A, Marino B, Carrozzo R, Korenberg JR, Danesino C, Sujanski E, Dallapiccola B, Zuffardi O. Deletion of a 5-cM region at chromosme 8p23 is associated with a spectrum of congenital heart defects. Circulation 2000;102:432-437.

1999

Digilio MC, Marino B, Toscano A, Giannotti A, Dalla piccola B. Atrioventricular canal defect without Down sindrome: A heterogenous malformation. Am J Med Genet 1999;85:140-146.

Digilio MC, Marino B, Bagolan P, Giannotti A, Dallapiccola B. Microdeletion 22q11 and oesophageal atresia. J Med Genet 1999;36:137-139.

Digilio MC, Marino B, Ammirati A, Borzaga U, Giannotti A, Dallapiccola B. Cardiac malformations in patients with Oral-facial-skeletal syndromes: Clinical similarities with hetrotaxia. Am J Med Genet 1999;84:350-356.

Digilio MC, Marino B, Giannico S, Giannotti A, Dalla piccola B. Atrioventricular canal defect and hypoplastic left heart syndrome as discordant heart defects in twins. Teratology 1999;60:206-208.

Digilio MC, Marino B, Giannotti A, Mingarelli R, Dallapiccola B. Guidelines for 22q11 deletion screening of patients with conotruncal defects. J Am Coll Cardiol 1999;33:1746-1747.

Digilio MC, Pacifico C, Tieri L, Marino B, Giannotti A, Dallapiccola B. Audiological findings in patients with microdeletion 22q11 (DiGeorge/velocardiofacial syndrome). Br J Audiol 1999;33:324-329.

Digilio MC, Marino B, Bevilacqua M, Mugolino AM, Giannotti A, Dalla piccola B. Genetic heterogeneity of isolated noncompaction of the left ventricular myocardium. Am J Med Genet 1999;85:90-91.

Marino B, **Digilio MC**, Toscano A, Giannotti A, Dalla piccola B. Congenital heart diseases in children with Noonan's syndrome: An expanded cardiac spectrum with high prevalence of atrioventricular canal. J Pediatr 1999;135:703-706.

Marino B, **Digilio MC**, Persiani M, Di Donato R, Toscano A, Giannotti A, Dallapiccola B. Deletion 22q11 in patients with interrupted aortic arch. Am J Cardiol 1999;84:360-361.

Amati F, Conti E, Novelli A, Bengala M, **Digilio MC**, Marino B, Giannotti A, Gabrielli O, Novelli G, Dalla piccola B. Atypical deletions suggest five 22q11.2 critical regions related to the DiGeorge/velo-cardio-facial sindrome. Eur J Hum Genet 1999;7:903-909.

Botta A, Novelli G, Mari A, Novelli A, Sabani M, Korenberg J, Osborne LR, **Digilio MC**, Giannotti A, Dallapiccola B. Detection of an atypical 7q11.23 deletion in Williams syndrome patients which does not include the STX1A and FZD3 genes. J Med Genet 1999;36:478-480.

1998

Digilio MC, Marino B, Banaudi E, Marasini M., Dallapiccola B. Familial recurrence of transposition of the great arteries . Lancet 1998;351:1661.

Digilio MC, Marino B, Canepa SA, Borzaga U, Giannotti A, Dallapiccola B. Congenital heart defect in sibs with discordant karyotypes. Am J Med Genet 1998; 80:160-162.

Digilio MC, Marino B, Picchio F, Prandstraller D, oscano A, Giannotti A, Dalla piccola B. Noonan syndrome and aortic coarctation. Am J Med Genet 1998;80:160-162.

1997

Digilio MC, Marino B, Giannotti A, Toscano A, Dallapiccola B. Recurrence risk figures for isolated tetralogy of Fallot after screening for 22q11 microdeletion. J Med Genet 1997;34:188-190.

Digilio MC, Giannotti A, Marino B, Guadagni AM, Orzatesi M, Dallapiccola B. Radial aplasia and chromosome 22q11 deletion. J Med Genet 1997;34:942-944.

Digilio MC, Marino B, Giannotti A, Dallapiccola B. Conotruncal heart defect/microphthalmia sindrome: delineation of an autosomal recessive syndrome. J Med Genet 1997;34:927-929.

Digilio MC, Marino B, Borzaga U, Giannotti A, Dallapiccola B. Intrafamilial variability of Pfeiffer-type cardiocranial sindrome. Am J Med Genet 1997;73:480-483.

Digilio MC, Marino B, Giannotti A, Dalla piccola B. The atrioventricular canal defect is the cngenital heart disease connecting short-rib polydactyly and oral-facial-digital syndromes. Am J Med Genet 1997;68:110-112.

1996

Digilio MC, Marino B, Giannoti A, Dallapiccola B. Orocardiodigital sindrome: an oral-facial-digital type II variant associated with atrioventricular canal. J Med Genet 1996;33:416-418.

Digilio MC, Marino B, Grazioli S, Agostino D, Giannotti A, Dallapiccola B. Comparison of occurrence of genetic syndromes in ventricular septal defect with pulmonary stenosis (classic tetralogy of fallot) versus ventricular septal defect with ulmonic aresia. Am J Crdiol 1996;77:1375-1376.

1995

Digilio MC, Giannoti A, Pagnotta G, Mingarelli R, Dallapiccola B. Joint dislocation and cerebral anomalies are consistently associated with ora-facial digital syndrome type IV. Clin Genet 1995;48:156-159.

Digilio MC, Marino B, Giannotti A, Dallapiccola B. Single arium, atrioventricular canal / postaxial hexadactyly indicatine Ellis-van Creveld syndrome. Hum Genet 1995;96:251-253.

Digilio MC, Marino B, Formigari R, Giannotti A. Maternal diabetes causing DiGeorge anomaly and enal agenesis. Am J Med Genet 1995;55:513-514.

1994

Digilio MC, Giannotti A, Floridia G, Uccellatore F, Mingarelli R, Danesino C, Dalla piccola B, Zuffardi O. Trisomy 8 syndrome owing to isodicentric 8p chromosomes: regional assignment of a presumptive gene involved in corpus callosum development. J Med Genet 1994;31:238-241.

Digilio MC, Mingarelli R, Marino B, Ginanotti A, Melchionda S, Dallapiccola B. Congenital cardiac defect in a patient with mosaic 45,X/46,XX,i(21q) karyotype. Clin Genet 1994;46:268-270.

Digilio MC, Marino B, Giannotti A, Dalla piccola B. Familial atrioventricular septal defect: possibile genetic mechanisms. Br Heart J 1994;72:301.

1993

Digilio MC, Marino B, Cicini MP, Giannotti A, Formigari R, Dallapiccola B. Risk of congenital heart defects in relatives of patients with arioventricular canal. Am J Dis Child 1993;147:1295-1297.

Digilio MC, Giannotti A, Marino B, Dallapiccola B. Atrioventricular canal and 8p-syndrome. Am J Med Genet 1993;47:437-438.

Digilio MC, Giannotti A, Marino B, Obregon MG, Dallapiccola B. Discrete membranous subaortic stenosis in siblings. Eur J Pediatr 1993;152:622.

1992

Marino B, Reale A, Giannotti A, **Digilio MC,** Dallapiccola B. Nonrandom association of atrioventricular canal and del(8p) syndrome. Am J Med Genet 1992;42:424-427.

Giannotti A, **Digilio MC**, Standoli L, Zama M, Dallapiccola B. A new case of Bartsocas-Papas syndrome surviving at 20 months. Am J Med Genet 1992;42:733-735.

Dallapiccola B, Giannotti A, Marino B, **Digilio MC**, Obregon MG. Familial aplasia cutis congenita and coarctation of the aorta. Am J Med Genet 1992;43:762-763.

1991

Digilio MC, Marino B, Giannotti A, Dallapiccola B. Trisomia 18 associata a canale atrioventricolare. G It Cardiol 1991;21:433-435.

Marino B, **Digilio MC**, Papa M, Giannotti A, Dallapiccola B. Turner's syndrome with atrioventricular canal. Pediatr Cardiol 1991;12:245-246.

1989

Marino B, **Digilio MC**, Giannotti A, Dallapiccola B. Atrioventricular canal associated with trisomy 9. Chest 1989;96:1420-1421.