

Table 2 Clinical features of the 14 newly described patients carrying NF1 microdeletion characterised by refined fluorescent in situ hybridisation (FISH) analysis

Patient	Age (years)	Sex	Deletion type	Clinical signs				
				Growth defects	Dysmorphic	Mental retardation	Cardiovascular malformation	Other features
119	4	M	REP	No	No	No	–	
118	5	M	cen-REP	No	No	No	–	Optic glioma, seizures
93	6	M	REP	90th centile, macrocephaly	Yes*	No	HCM	Broad neck, 3 NFs
65	6	M	REP	Height 3rd centile, microcephaly 2nd centile	Yes†	IQ48	VSD (upper part)	Small hands/feet, short fingers
116	6	M	REP	Short stature 10th centile	Yes‡	IQ54	Mitral insufficiency	MCLS, kyphoscoliosis, pectus excavatum, genu valgus, pes planus, umbilical hernia
72	7	M	REP	No	Yes§	IQ50	–	Small hands/feet, short fingers
76	8	F	REP	No	Yes¶	No	–	–
94	8	F	REP	No	Yes**	No	–	–
75	9	F	REP	No	Yes††	No	–	–
85	11	M	REP	No	No	IQ77	–	MCLS
7	11	M	REP	No	Yes‡‡	Speech impairment	–	MCLS, amblyopia, thalamic hamartoma
82	23	F	REP	Short stature	No	No	–	Hearing impairment, Noonan-like
77	U	F	REP	Overgrowth >97th centile	Yes§§	Speech impairment, LD	–	NFs, required special education, short and broad fingers and toes
78	U	F	REP	–	Yes¶¶	Speech impairment	–	Delayed motor development, short and broad feet, fifth finger clinodactyly

*Hypertelorism, epicanthic folds, low set ears, low posterior hairline.

†Hypertelorism, downslanting eye, strabismus, large and prominent nose with high and broad bridge, bulbous nasal tip, large and low set ears, malar hypoplasia, wide and prominent philtrum, small mouth, small pointed chin.

‡Hypertelorism, long philtrum, broad nose.

§Prominent forehead, hypertelorism, ptosis (O.DX), downslanting eyes; strabismus, large and prominent nose with high and broad bridge and bulbous nasal tip, large and low set ears, wide and prominent philtrum, low posterior hairline.

¶Coarse face, hypertelorism.

**††Epicanthic folds.

‡‡Hypertelorism, broad and wild nasal bridge, broad nasal tip.

§§Simple facial features.

¶¶Epicanthic folds, bulbous nose, narrow high palate, low forehead.

cen-REP, deletion extending centromerically to REP-P; CVM, cardiovascular malformations; F, female; HCM, hypertrophic cardiomyopathy; LD, learning disabilities; M, male; MCLS, multiple café-au-lait spots; NF, neurofibroma; REP, NF1 repeat mediated deletion; U, unknown; VSD, ventricular septal defect.

Key points

- *NF1* microdeletion syndrome is determined by haploinsufficiency of the *NF1* gene and its flanking regions; *NF1* microdeleted patients show a more severe phenotype than observed in classical *NF1* patients.
- The aim of this study was to verify the presence of specific clinical signs of *NF1* microdeletion, by combining clinical and genetic evidence from 92 deleted patients, 14 newly characterised and 78 already published.
- Statistical analysis, done by comparing the frequency of 10 clinical signs between *NF1* microdeleted patients and the whole *NF1* population, showed that the most common extra-*NF1* clinical signs in microdeleted patients were mental retardation, cardiovascular malformations, and dysmorphisms.
- Using bioinformatic tools, the deletion gene content of 44 genetically and clinically characterised patients was established. It is proposed that haploinsufficiency of *OMG* and/or *CDK5R1* genes may be implicated in mental retardation. In relation to cardiovascular malformations, only *JJAZ1* and *CENTA2* can be considered as plausible candidate genes.
- When present in an *NF1* patient, dysmorphisms, cardiac anomalies, and mental retardation are signs indicating *NF1* microdeletion.

CORRECTION

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Owing to the change over of typesetters at the BMJ Publishing Group Ltd last year, we would like to apologise for an error that was published in the paper by Baser *et al* (*J Med Genet* 2003;40:758–760). In Table 2 the headings ‘Classical NF2’ and ‘Somatic mosaic’ should be indented as they are subsections of the heading ‘Nonsense or frame-shift’.